Oesophagus and Stomach

Systems: Digestive

Causes: Genetic, blood supply, infection, cancer

Introduction

The diseases of systems are divided into congenital and acquired. The acquired are then divided into non-neoplastic and neoplastic.

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OESOPHAGUS
Congenital

Oesophageal Atresia

Definition: Esophageal atresia is a serious birth defect in which the esophagus, that connects the mouth to the stomach, is segmented and closed off at any point. This condition usually occurs with a condition in which the esophagus is improperly attached to the trachea.

Sites:

During fetal development, the esophagus and the trachea arise from the same original tissue. Normally, the two tubes would form separately; however, in cases of esophageal atresia and tracheoesophageal fistulas, they do not. The most common configuration is the type c, - see below - in which the upper part of the esophagus abruptly ends in a blind pouch, while the lower part attaches itself to the trachea. This configuration occurs in 85-90% of cases. Esophageal atresia without involvement of the trachea occurs in only 8% of cases.

Incidence: Esophageal atresia occurs in approximately 1 in 4,000 live births.

Peak age: at birth. 30% are born prematurely

Gender: No convincing gender or racial preponderance

Other demographics: nil

Clinical Presentation: An infant born with this defect will at first appear normal, swallowing normally. However, the blind pouch will begin to fill with mucus and saliva that would normally pass through the esophagus to the stomach, causing the baby to drool excessively. When fed, the baby will also immediately regurgitate what he or she has eaten. Choking and coughing may also occur as the baby...
breaths in the fluid overflowing from the esophagus. Aspiration pneumonia, caused by inhalation of the contents of the digestive tract, may also develop.

**Molecular pathogenesis: Chromosomal Causes**

Chromosome anomalies have been reported in approximately 6%-10% of individuals with oesophageal atresia/tracheo-oesophageal fistula (EA/TEF).

EA/TEF is found in the following aneuploidy syndromes. Aneuploidy is the state of having chromosomes in a number that is not an exact multiple of the haploid number.

- Trisomy 21 (~0.5%-1.0% of affected individuals)
- Trisomy 18 (~25% of affected individuals)
- Trisomy 13

**Risk Factors:** none known

**Macroscopic appearance:** see imaging

**Imaging:**

*Pre-natal* EA/TEF may be suspected if ultrasound examination reveals polyhydramnios, absence of a fluid-filled stomach, a small abdomen, lower-than-expected fetal weight, and a distended esophageal pouch. Foetal MRI may be used for final confirmation.

*Post-natal*

Doctors who suspect esophageal atresia diagnose the condition using x-ray imaging.

Plain xray of the abdomen will show absence of the gastric bubble. A nasogastric tube will curl up in the blind pouch and can be seen on plain xray of the chest.

A small amount of contrast can be injected down the tube that has stopped a few cms down the oesophagus, to confirm the anatomy.

**Microscopic appearance:** The oesophagus and trachea develop from the same tube so the oesophagus starts with a lining of columnar epithelium which later becomes stratified squamous epithelium.
Biochemistry: Nil

Diagnosis: by passing a catheter through the nose and into the esophagus. Esophageal atresia is indicated if the catheter hits an obstruction 10-13 cm from the nostrils.

Treatment:

Infants with esophageal atresia are unlikely to survive without surgery to reconnect the esophagus. The procedure is done as soon as possible after birth. Prematurity, the presence of other birth defects, or complications of aspiration pneumonia may delay surgery. Once diagnosed, the baby will be fed intravenously until he or she has recovered sufficiently from the operation. Mucus and saliva will also be continuously removed via a catheter until recovery has occurred. When surgery is performed, the esophagus is reconnected and, if necessary, separated from the trachea. If the two ends of the esophagus are too far apart to be reattached, tissue from the large intestine is used to join them.

Prognosis: Surgery to correct esophageal atresia is usually successful. Post-operative complications may include difficulty swallowing, since the esophagus may not contract efficiently. Gastro-oesophageal reflux, in which the acidic contents of stomach flow back into the oesophagus, can cause ulcers.

Right sided Aortic Arch compressing the Oesophagus

Incidence: 0.1% of the population.


Angiogram Courtesy Dr Maxime St-Armand. Radiopaedia.org, rID 20698 – see arrow where the anomalous left subclavian artery passes behind the oesophagus, causing compression and dysphagia.

A barium swallow shows the indentation of the left subclavian artery upon the posterior wall of the oesophagus – see arrow. Image courtesy of Radiology in Pediatric Emergency Medicine. Vol 6, Case 20.
Pulmonary sling occurs because of failure of formation of the left 6th aortic arch so there is absence of the left pulmonary artery. The blood to the left lung comes from an aberrant left pulmonary artery which arises from the right pulmonary artery and crosses between the oesophagus and trachea, compressing the oesophagus from its anterior surface – see arrow. Image courtesy of Learning Radiology 2010.

Acquired Conditions of the Oesophagus

Non-neoplastic

Foreign bodies
**Types:** chicken bones, coins and small toys are opaque to x-rays but fish bones are non-opaque.

**Site:** 70% of impactions occur just below the cricopharyngeous. 20% occur at the level of the aortic arch and 10% at the oesophago-gastric junction.

**Clinical presentation:** dysphagia (difficulty in swallowing; food feels stuck in the chest or throat).

**Diagnosis:** plain film x-rays are obtained first but often will not demonstrate the foreign body except coins. Image, courtesy of LearningRadiology.com. V.Rooks and E.Chung, shows a chicken bone lodged in the oesophagus – see arrow.

A gastrographin or thin barium swallow may then be used.

**Oesophageal diverticulae**

**Incidence:** 0.1% of the population.

**Gender:** more common in males

**Site:** Cervical region, thoracic and oesophago-gastric junction.

**True:** all the layers of the wall remain present e.g. Killian-Jamieson.

**False:** no muscle layer exists e.g. Zenker’s diverticulum. Sometimes called pseudodiverticulae.

Also false diverticulae caused by surgical trauma.

**Macroscopic appearance:**
Zenker's diverticulum: *this is directed posteriorly* in the midline through the cricopharyngeus muscle within the hypopharynx. i.e. these arise from the hypopharynx, rather than the oesophagus. Presents in patients over the age of 50 years.

**Micro:** Zenker diverticula are lined with stratified squamous epithelium with a thin lamina propria. No muscular layer exists. Fibrosis surrounding the diverticulum is common.

In the image of a Zenker’s diverticulum below, courtesy of Dr J Heilman. Wikimedia Commons, see a bilobed diverticulum. Typical level is C5/C6 disc level.

![Image of Zenker's diverticulum](https://commons.wikimedia.org/wiki/File:Diverticulum_of_the_hypopharynx.jpg)

90% of Zenker diverticulum also have a hiatus hernia and gastro-oesophageal reflux.

**Kilian-Jamieson:** *this is directed anterior and laterally* just below the cricopharyngeus. It is often left sided but can be bilateral. Much smaller than a Zenker’s diverticulum and also seen less frequently. Also infrequently symptomatic.

Clinical Presentation:  dysphagia, halitosis, regurgitation of undigested food, a sensation of a lump in the throat.

Age:  middle aged adults and the elderly.

Treatment:  diverticulectomy and myomectomy.

Prognosis:  if left untreated both the Zenker’s diverticulum and the Killian-Jamieson diverticulum have the complication of aspiration.

**Oesophageal Varices**
**Definition:** These are extremely dilated sub-mucosal veins, usually in the lower third of the oesophagus but may in superior vena caval obstruction, involve the entire oesophagus.

**Etiology:** most often a consequence of portal hypertension due to cirrhosis of the liver.

**Clinical presentation:** may be completely silent until present with catastrophic bleeding when one of the varices ruptures.

**Diagnosis:** Esophageal varices are diagnosed with a barium swallow imaging the oesophagus and/or by endoscopy.

**Macroscopic appearance:**

![Image of esophageal varices](image_url)

**Radiology:**

Image below, courtesy of J Carale. Medscape article 182098 2014. This is a barium swallow, and shows two projections. The varices have involved the entire oesophagus – see arrows.
Endoscopy of same patient showing dilated veins – see arrow – and some red bleeding points.

Pathogenesis: When the portal vein or its intrahepatic branches is obstructed, in order for blood to return to the heart, an ancillary route has to open up. Back pressure drives blood to the coronary veins of the stomach and then into the mucosal and submucosal veins of the oesophagus. From the middle one third, blood drains to the azygos veins and is then returned to the systemic circulation via the superior vena cava from the upper one third of the oesophagus. These are called uphill varices.

Two thirds of the patients with cirrhosis, often due to alcoholism, will develop varices.

However, in patients who have an obstructed superior vena cava (SVC), as for example by carcinoma of the lung, blood can only return to the heart from the head, neck and upper limbs via collateral channels in the oesophagus so pressure builds up producing downhill varices.

If the SVC is obstructed above the level of the azygos vein, the varices are confined to the upper and mid thoracic oesophagus.
If the SVC obstruction is below the azygos vein, blood cannot use the azygos vein so varices involve the entire thoracic oesophagus, with blood returning to the heart via the inferior vena cava.

**Treatment:** a temporary tamponade with a Sangstaken tube balloon can be achieved and then surgery to band the varices or inject sclerosing agents.

**Prognosis:** 40% die with the initial bleed.

Rebleed in survivors occurs within one year, and again 40% of these will die.

**Motor disorders**

**Achalasia (cardiospasm)**

Occurs as secondary achalasia in Chagas Disease (trypanosomiasis) which is found in South America Known as American trypanosomiasis, is a tropical parasitic disease *Trypanosoma cruzi* and spread mostly by insects known as Triatominae or kissing bugs because they tend to bite on the face.

Image courtesy of Wikimedia Commons

The disease can also be transmitted via blood transfusions.

Damage to the heart, brain, intestine and oesophagus is occurring during the infective cycle, by sequentially inducing an inflammatory response, cellular lesions and fibrosis.

**Treatment:** Antiparasitic treatment is most effective early in the course of infection, but is not limited to cases in the acute phase. Drugs of choice include azole or nitro derivatives, such as bensnidazole or nifurtimox. Both agents are limited in their capacity to completely eliminate *T. cruzi* from the body (parasitologic cure), especially in chronically infected patients, and resistance to these drugs has been reported.

**Prognosis:** is supportive. Dilatation of the fibrosed oesophagus can occasionally result in perforation and mediastinitis.

**Scleroderma**
**Definition:** Scleroderma is an autoimmune disease which is progressive that affects the skin and connective tissue (including cartilage, bone, fat, and the tissue that supports the nerves and blood vessels throughout the body) by depositing excessive amounts of collagen. There are two major forms of the disorder. The type known as localized scleroderma mainly affects the skin. Systemic scleroderma, also called systemic sclerosis, affects the smaller blood vessels and internal organs of the body, including the oesophagus.

**Clinical:** The oesophagus is involved in 80% of cases of scleroderma and becomes stiff and scarred. Patients may have trouble swallowing food. The acid contents of the stomach may start to flow into the esophagus (esophageal reflux).

**X-ray appearance:** The barium swallow will show no peristalsis in the oesophagus which appears dilated and atonic and the lower oesophageal sphincter is wide open so the patient suffers ‘heart burn’ from reflux of gastric acid content. Image courtesy of Flashcards. Rheum #5 2012.

**Prognosis:** 65% of patients survive 11 years following diagnosis.

**Plummer-Vinson syndrome**

**Definition:** there is abnormal oesophageal peristalsis coupled with oesophageal strictures due to fibrosis in the post-cricoid region. There are also projections of mucosal webs into the lumen.

**Incidence:** now rare because nutrition has been vastly improved in the last 50 years.

**Age:** 40 – 70 years

**Gender:** 90% are women
Clinical presentation: post-cricoid dysphagia, upper oesophageal webs and iron deficiency anaemia triad comprises the syndrome. Patients complain of a burning sensation on the tongue and oral mucosa, and have atrophy of lingual papillae producing a smooth, shiny, red, dorsum of the tongue.

Risk factors: Patients also have an increased incidence of carcinoma of the tongue and oesophagus.

X-ray appearance:

Image below is a barium swallow in a patient with Plummer-Vinson syndrome showing the multiple sites of stricture. Fluoroscopy would show disorganised peristalsis.

Microscopic appearance: a few studies have been postmortem and showed hyperkeratinization and partial atrophy of the epithelium with degeneration and atrophic changes in the underlying muscle of the oesophagus.

Treatment: correct the iron deficiency anaemia which improves the dysphagia and the pain.

If the web can be dilated, using endoscopy, this will improve dysphagia and restore normal swallowing.

Prognosis: respond well to treatment with iron supplements resolving the anaemia and improving the pain on the tongue.

Infective Oesophagitis

Causative Agents: candida, herpes simplex, cytomegalic inclusion disease, HIV.

Clinical presentation: may be asymptomatic, but typical symptoms include the following: Onset of difficult or painful swallowing (ie, dysphagia or odynophagia)

- Heartburn
• Nausea
• Fever
• Epigastric pain
• Anorexia
• Cough

Retrosternal discomfort or pain and vomiting and sepsis
Abdominal pain
Hematemesis and weight loss

X-ray appearance: Images courtesy of D. Devuni Medscape 2015

Barium Swallow of a patient with *Candida oesophagitis*. There are linear vertical plaque like lesions with normal intervening mucosa.

Barium swallow below is *herpes simplex oesophagitis* which shows multiple ulcers – see arrows.

Image below is a Barium swallow of a patient with *cytomegalic inclusion disease oesophagitis*. Note the large flat ulcer – big arrows and a cluster of small ulcers – small arrows. The patient also had AIDS. As HIV oesophagitis looks the same as cytomegalic inclusion disease oesophagitis, the patient needs to have an endoscopy to obtain a sample before commencing treatment.
Barrett oesophagus – premalignant condition

**Definition:** Barrett esophagus is defined as displacement of the squamocolumnar junction proximal to the gastroesophageal junction with histological evidence of specialized intestinal metaplasia on biopsy specimens.

This is a complication of reflux of acid contents from the stomach into the oesophagus and consists of the stratified squamous epithelium mucosa in the oesophagus being replaced by metaplastic columnar epithelium. The intestinal metaplasia includes the presence of goblet cells.

**Incidence:** about 10% of patients with gastro-esophageal reflux develop Barrett’s oesophagus.

**Peak age:** over the age of 50 years.

**Gender:** more common in males

**Risk factors:** obesity because this increases the chance of gastric reflux.

**Diagnosis:** endoscopy and biopsy – see image below courtesy of Sharma, P. et al. The development and validation of an endoscopic grading system for Barrett’s esophagus: the Prague C & M criteria. Gastroenterology 131, 1392-1399 (2006).

*The Prague C (circumferential) and M (maximal extent) criteria* were developed and validated by Sharma et al. in 2006. In this classification, both the maximal length (M) (including tongues) of Barrett esophagus, as well as the length of the circumferential Barrett segment (C) are measured during endoscopy. These numbers can then be used to track the length of the Barrett segment over time.
Microscopic appearance:

Image courtesy of Dr M R Wick, PathologyOutlines.com April 2013. Black arrow indicates a goblet cell and the white arrow points to a gastric type columnar cell.

Treatment: Medical measures to prevent gastric reflux such as losing weight and sleeping sitting up.

Prognosis: Less than 1% of those with Barrett’s oesophagus develop cancer of the oesophagus.

If the segment of changed epithelium exceeds 2 cm, there is a 40 times increase in the incidence of adenocarcinoma. For this reason Endoscopy every 1-2 years to detect dysplasia or early adenocarcinoma with 4 quadrant biopsies is recommended in those with Barrett’s oesophagus.

Carcinoma of the Oesophagus

Definition:

Esophageal carcinomas arise from the epithelial lining of the esophagus and fall into one of two classes:
esophageal squamous-cell carcinomas ESCC, which are similar to head and neck cancers in their appearance

and esophageal adenocarcinomas (EAC), which are often associated with a history of gastro-oesophageal reflux disorder and Barrett's esophagus.

Sites:
A cancer in the upper two-thirds is likely to be ESCC and one in the lower one-third EAC.

Squamous cell carcinoma accounts for 90% of cases. 20% occur in the upper one third, 50% in the middle third and 30% in the lower third.

Incidence: 8th most common cancer globally. EAC is 0.7 per 100,000 ESCC is 5.0 per 100,000

Peak Age: 83% are over the age of 60 years

Gender: M : F = 3 : 1 for ESCC and M : F = 10 : 1 for EAC.

Other demographics: For ESCC, the condition is 6 times more common in African-Americans than in Caucasians.

Clinical presentation: Significant symptoms usually do not appear until the cancer has infiltrated over 60% of the circumference of the esophageal wall, by which time the tumor is already in an advanced stage. Onset of symptoms is usually caused by narrowing of the lumen due to the physical presence of the tumour.

The first and the most common symptom is usually difficulty in swallowing.

Pain behind the sternum may be severe, made worse by swallowing foodstuffs.

Patient is at risk of aspiration pneumonia partly due to fistulae that may occur between the oesophageal tumour and the trachea.

Molecular pathogenesis: for ESCC it is not completely known but loss of several tumour suppressor genes, including p53 and p16/INK4a is involved.

Risk factors: ESCC association with tobacco and alcohol consumption—more common in the developing world.

EAC: More common in the developed world – association with gastro-oesophageal reflux.

Macroscopic appearance: ESCC – 60% are polypoid, 15% diffusely infiltrating and 25% ulcerated.
X-ray:

Barium Swallow and meal:

*Squamous cell carcinoma*: courtesy of Dr F Gaillard. Radiopaedia.org, rID4232. The image demonstrates a polypoid type and the 3 images labelled with time intervals show how the narrowing of the lumen is causing considerable hold-up of barium above it, even after 5 minutes. From that situation, a patient may aspirate, especially if they sleep lying flat.
Image courtesy Dr A Stanislavsky. Radiopaedia.org, rID12917 displays an adenocarcinoma with a ragged luminal surface suggestive of the ulcerating type of ESCC.

**CT scan** is undertaken to assess the spread in the mediastinum and at the same examination identify any metastases in the lungs, neck and abdomen.

When the carcinoma spreads laterally to the right, it can encircle and damage the right recurrent laryngeal nerve which results in the patient having a hoarse voice.

The image below, courtesy of Dr P K Desai, shows the lateral spread of the tumour in such a case.
Microscopic appearance:

**Squamous cell carcinoma:**

Image below courtesy of Dr E Weisenberg. PathologyOutlines.com 2014

The normal squamous epithelium at the left – see arrow, merges into the squamous cell carcinoma at the right – see block arrow, which is infiltrating downward. The neoplastic squamous cells are still similar to the normal squamous cells, but are less orderly. This is a well-differentiated squamous cell carcinoma, because the neoplastic cells still have many features of the squamous cell of origin.

**Adenocarcinoma:** image courtesy of Wikimedia Commons
Spread: is via the lymphatics. As the thoracic oesophagus does not have a serosa covering it, only loose connective tissue called adventitia, there is not a firm covering to limit the spread of cancer.

Staging

is based on the TNM staging system, which classifies the amount of tumor invasion (T), involvement of lymph nodes(N), and distant metastasis (M). The currently preferred classification is the 2010 AJCC staging system (American Joint Committee on Cancer) for cancer of the esophagus and the esophagogastric junction and this system also incorporates information on cell type (ESCC, EAC, etc.), grade (degree of differentiation – an indication of the biological aggressiveness of the cancer cells), and tumor location (upper, middle, lower, or junctional).

Biochemistry: nil

Diagnosis: endoscopy and biopsy and imaging techniques such as CT scan and MRI

Treatment: treatment aiming at cure is restricted to localized disease, without distant metastasis: in such cases a combined approach that includes surgery may be considered.

Disease that is widespread, metastatic or recurrent is managed with palliation. The tumour may be responsive to chemotherapy which can lengthen survival, while treatments such as radiotherapy or stenting may be used to relieve symptoms and make it easier to swallow.

Image courtesy of Dr J Jones. Radiopaedia.org, rID 8815 shows the outline of a stent projected over the mediastinal portion of the heart – see arrows.
**Prognosis:** 5-year survival is 20%.

**Prevention:** stop smoking and excessive alcohol consumption for ESCC

Uncommon oesophageal tumours includes;

Carcinoid, lymphoma, melanoma, leiomyosarcoma and rhabdomyosarcoma.

**STOMACH**

**Congenital**

**Infantile Hypertrophic Pyloric Stenosis**

**Definition:** Pyloric stenosis refers to a narrowing of the passage between the stomach and the small intestine. Occurs secondary to hypertrophy and hyperplasia of the muscular layers of the pylorus, causing a functional gastric outlet obstruction.

Marked hypertrophy and hyperplasia of the 2 (circular and longitudinal) muscular layers of the pylorus occurs, leading to narrowing of the gastric antrum. The pyloric canal becomes lengthened, and the whole pylorus becomes thickened. The mucosa is usually edematous and thickened. In advanced cases, the stomach becomes markedly dilated in response to near-complete obstruction.

**Incidence:** 1 in 4000 babies.

**Age:** develops 20 - 40 days after birth. Range is one day to 5 months. Infantile hypertrophic pyloric stenosis is rare in premature infants.

**Gender:** M : F =4 : 1. Female babies tend to present with symptoms later than male babies.
Etiology: The causes of infantile hypertrophic pyloric stenosis are multifactorial. Both environmental factors and hereditary factors are believed to be contributory. Possible etiologic factors include deficiency of nitric oxide synthase containing neurons, abnormal myenteric plexus innervation, infantile hypergastrinemia, and exposure to macrolide antibiotics. It may be inherited in some cases, as several members of a family may have had the condition in infancy.

Clinical Presentation: forceful and frequent vomiting. As the baby becomes dehydrated, the anterior fontanelle and the eyes appear sunken. Constipation is evident as no food is reaching the intestine. Urination may only occur 4 – 6 hours apart. Salt as well as fluid imbalance occurs. Baby does not gain weight or may even lose weight.

Diagnosis: the muscular mass can be felt by palpation of the abdomen. If this is not found, an ultrasound can confirm the diagnosis or a barium swallow.

Macroscopic appearance: muscles in the pylorus have enlarged preventing food emptying. The diagram and the appearance at surgery are courtesy of Dr M Hernanz-Schulman RSNA Radiology 227(2) 2003.

X-ray appearance: Ultrasound is the preferred method. Image below courtesy of Dr F Gaillard. Radiopaedia.org, rID 8142. M = hypertrophied muscle of the pylorus.
Microscopic:

Microscopically, circular muscle hypertrophies, with increased connective tissue in the septa between the muscle bundles. An increase in chondroitin sulphate within the extracellular matrix may account for the cartilaginous quality of the pyloric tumor. Note that a histologic specimen is not obtained at operation nor is it necessary for the diagnosis of pyloric stenosis.

Confocal laser microscopy producing a 3-dimensional view, is able to identify abnormally thick contorted nerve bundles in the pyloric muscle layers of infants with IHPS. The etiology of infantile hypertrophic pyloric stenosis may be related to these abnormal nerve fibers.


Treatment: Pyloric stenosis can be cured with a surgical procedure called a pyloromyotomy. In this operation, the surgeon makes an incision in the baby's abdomen. Then a small cut is made in the thickened muscle of the pylorus and it is spread apart. So the passage can be widened without removing any tissue. (The procedure may be performed with the aid of a laparoscope.) After surgery, the pylorus will heal itself. The thickening gradually goes away and the passage resumes a normal shape.

Prognosis: Pyloromyotomy that is adequately performed is curative of pyloric stenosis. There have been reports of recurrent pyloric stenosis despite performance of an adequate pyloromyotomy, but recurrence is considered to be a rare exception after incomplete pyloromyotomy has been ruled out.

After pyloromyotomy, the pylorus changes significantly within 3 days postoperatively and returns to normal within 5 months.


Congenital diaphragmatic hernias

Bochdalek hernia

it is the most common manifestation of congenital diaphragmatic hernia, accounting for more than 95% of cases.

Definition: the diaphragm abnormality is characterized by a hole in the postero-lateral corner of the diaphragm which allows passage of the abdominal viscera into the chest cavity. 85% occur on the left side of the diaphragm, a large proportion of the remaining cases occur on the right.

Incidence: rare.

Clinical presentation: In the infant, it can be a cause of pulmonary hypoplasia and of respiratory distress. Adults may have asymptomatic hernias which are identified when imaging is for another purpose.

X-ray: the finding can be visible on a plain chest radiograph as in the image below, courtesy of M.Tan AJR 2001, 177:363-366.
It is also possible to diagnose the hernia in utero – see image below

Treatment: the colon or stomach is pulled back through the hole into the abdominal cavity and the hole in the diaphragm repaired.

**Morgagni Hernia**

**Definition:** this is due to an anterior defect of the diaphragm and is characterized by herniation of abdominal organs through the foramina of Morgagni which are located immediately adjacent to the xiphoid process of the sternum.

**Frequency:** accounts for only 2% of all congenital diaphragmatic herniae.

**X-ray appearance:** the lateral view of the chest and the lateral view of a barium study images are courtesy of Drs P Mudgal and J Jones. Radiopaedia 2014.
Treatment: is surgical repair as patients get respiratory distress and chest infections. Also bowel can prolapse and strangulate.

Diaphragm Eventration

Definition: congenital diaphragmatic eventration is when there is elevation of part or all of an otherwise intact leaf of the hemi-diaphragm into the chest cavity.

Etiology: in the region of the eventration the diaphragm is composed of fibrous tissue with only a few muscle fibres, allowing the abdominal viscera to protrude upwards.

It can occur as an acquired condition when there is phrenic nerve paralysis.

Xray appearance: the bulge is apparent on a chest radiograph. The significance can be checked by the radiologist observing the patient breathing via fluoroscopy and asking them to sniff. When there is an eventration as the cause of the bulge in the diaphragm, that part of the diaphragm does not move.

Situs Inversus
**Definition:**

is a congenital condition in which the major visceral organs are reversed or mirrored from their normal positions. Although cardiac problems are more common than in the general population, most people with situs inversus have no medical symptoms or complications resulting from the condition, and until the advent of modern medicine it was usually undiagnosed.

There is a 10% prevalence of congenital heart disease in situs inversus totalis and it is usually transposition of the great vessels.

**Genetic:** Situs inversus is generally an autosomal recessive genetic condition, although it can be X-linked.

**Incidence:** 1 in 10,000.

About 25% of individuals with situs inversus have an underlying condition known as primary ciliary dyskinesia (PCD). PCD is a dysfunction of the cilia that manifests itself during the embryologic phase of development. Normally functioning cilia determine the position of the internal organs during early embryological development, and so embryos with PCD have a 50% chance of developing situs inversus. The result is Kartagener syndrome, characterized by the triad of situs inversus, chronic sinusitis, and bronchiectasis. Cilia are also responsible for clearing mucus from the lung, and the dysfunction causes increased susceptibility to lung infections. Kartagener syndrome can also manifest with male infertility as functional cilia are required for proper sperm flagella function. Images from Wikipedia
Situs ambiguous or heterotaxy,

This is much rarer and the situs cannot be determined. In these patients, the liver may be midline, the spleen absent or multiple, and the bowel malrotated. Often structures are duplicated or absent altogether. This is more likely to cause medical problems than situs inversus totalis.

Acquired Conditions

Non-neoplastic

Hiatus Hernia

**Definition:** part of the stomach herniates into the thoracic cavity through the oesophageal hiatus in the diaphragm.

**Sites:** 95% are of the sliding type where the gastro-oesophageal junction is above the diaphragm.

5% are the rolling or paraoesophageal type when the gastro-oesophageal junction is below the diaphragm in its usual position.

**Association:** with gastro-oesophageal reflux which results in oesophagitis.

**Incidence:** as many are asymptomatic, this figure is unknown.

**Peak age:** over 50’s

**Gender:** more common in women.

**Other demographics:** nil

**Causes:** injury to the diaphragmatic hiatus for the oesophagus, persistent pressure on surrounding muscles e.g. coughing, constipation and lifting heavy objects.

**Risk factors:** being over 50 years, obese and pregnancy.

**Clinical presentation:** heartburn, belching, chest or abdominal pain, feeling especially full after meals and occasionally vomiting blood or passing black stools indicating gastrointestinal bleeding.

**Molecular pathogenesis:** nil

**Macroscopic appearance:** In the diagram below, courtesy of Dr W A Qureshi. Medscape Oct 30 2014.

1. is the normal state, 2. is the sliding type and 3. is the para-oesophageal.
X-ray appearance:

**Sliding type:** Chest xray, courtesy of Radiopaedia.org, rID 11493 and barium study courtesy of Dr J. Jones. Radiopaedia.org, rID 613

**Rolling type:** Chest xray courtesy of Radiopaedia.org, rID 11494 and the CT courtesy of Dr Petrali Carlo Lucius E. Radiopaedia.org, rID 11081.
Sometimes a hiatus hernia is not obvious on the PA chest x-ray as the heart is superimposed upon it. A lateral chest x-ray should always be included. See image below, courtesy of Dr A A Rabour. Radiopaemia.org, rID 24355.

One complication of a hiatus hernia is that the portion within the chest can rotate on its long axis causing a volvulus of the stomach. This is an emergency.

Image below courtesy of Dr D Cuete. Radiopaedia.org, rID 24355.
Microscopic appearance: nil
Biochemistry: nil
Diagnosis: barium swallow, CT scan and endoscopy
Treatment:

- Antacids that neutralize stomach acid.
- Medications to reduce acid production. H-2-receptor blockers, these medications include cimetidine and ranitidine
- Medications that block acid production and heal the esophagus. Proton pump inhibitors block acid production and allow time for damaged esophageal tissue to heal.
- surgery

Prognosis: variable but surgery can be curative.

**Volvulus of the Stomach**

There are two types of volvulus and both are a surgical emergency.

Images below are courtesy of A.Goel and J Jones et al. Radiopaedia 2014.

**Organo-axial volvulus** is when there is a large sliding hiatus hernia in the chest and it has twisted on its long axis. The outcome will be compromise of the blood supply with necrosis and gangrene.
Mesentero-axial volvulus is when the stomach rotates on the short axis from the lesser to the greater curve. Antrum and pylorus are above the fundus and body of the stomach.

Infection – Gastritis

Acute gastritis: this is an immediate response to noxious stimuli such as excessive alcohol consumption or the ingestion of corrosive agents. It can also occur as a response to severe renal failure.

Chronic gastritis:

Etiology: 90% have infection by Helicobacter pylori (Marshall and Warren). This is a gram negative rod and produces a urease which in turn produces ammonia which can be detected on the patient’s breath.

Only 15% of those infected with H.pylori proceed to develop gastric ulceration.

10% of cases of chronic gastritis are on an autoimmune basis. The patient loses the ability to produce intrinsic factor and will develop pernicious anaemia. This can co-exist with other autoimmune conditions such as Hashimoto’s thyroiditis and Addison’s disease of the adrenal glands.

Diagnosis: Urease breath test is the simplest.
Patient blows into a sealed tube containing $^{13}\text{C}$ which will produce $^{13}\text{CO}_2$ as a baseline. They are then given a drink containing $^{13}\text{C}$-urea, citric acid, aspartame, mannitol. H.pylori will break down the substrate as it produces urease. Then $^{13}\text{CO}_2$ is absorbed into the bloodstream and can be exhaled in the breath.

30 minutes later the patient blows into a second sealed tube and produces $^{13}\text{CO}_2$ plus ammonia. The two tubes are read by an isotope ratio mass spectrometer. The pre-drink and post-drink $^{13}\text{CO}_2$ should be the same if H.pylori is not present.

**Peptic Ulceration**

**Definition:** it is a break in the mucosa which extends through the muscularis mucosa into the submucosa or even deeper into adjacent organs.

**Sites:**
- first part of the duodenum – 73% of cases
- antrum of the stomach – 25%
- gastro-oesophageal junction
- at the margins of a gastrojejunostomy
- in a Meckel diverticulum which contains ectopic gastric mucosa
- multiple in the duodenum, stomach, jejunum in Zollinger-Ellison syndrome. In this a tumour secretes gastrin causing gastric acid production. Patients suffer diarrhoea and fatty stools. 25% are a part of MEN type 1, so will also have pituitary and parathyroid tumours.
- 20% of patients with gastric ulcer also have a duodenal ulcer.

**Etiology:** Helicobacter pylori organism, non-steroidal anti-inflammatory drugs.

**Also Stress Ulcers:**- *Curling ulcers* – occur in burns, trauma and in the proximal duodenum.

*Cushing’s ulcers* – occur in head injuries, post-craniotomy and often perforate.

**Incidence:** 4 / 1000 duodenal ulcers and 1 / 1000 gastric ulcers.

**Peak age:** 56 years

**Gender:** Men account for 58% of the duodenal ulcers and 52% of the gastric ulcers.

Women account for 42% of the duodenal ulcers and 48% of the gastric ulcers.

**Clinical presentation:** The most common symptoms are waking at night with upper abdominal pain or upper abdominal pain that improves with eating. The pain is often described as a burning or dull ache. Other symptoms include belching, vomiting, weight loss, or poor appetite. About a third of older people have no symptoms- Complications may include bleeding, perforation, and bleeding occurs in as many as 15% of people.
Molecular pathogenesis: nil.

Risk factors:  
I. Co-existence of other diseases:
   - Chronic renal failure
   - Zollinger-Ellison syndrome
   - $\alpha$-1 antitrypsin deficiency – one third have gastric ulcers and the figure is higher in those who also have lung disease.
   - Chronic lung disease – 25% have ulcers.

II. External Factors
   - Drugs e.g. aspirin, non-steroidal anti-inflammatory drugs
   - Cigarettes

Macroscopic appearance: Acute gastric ulcer
**X-ray appearance:** the barium study below shows the arrow pointing to an ulcer crater.

![X-ray Image](image1)

**Microscopic appearance:** images courtesy of Robbins and Cotran. Atlas of Pathology. 2nd ed

**Acute gastric ulcer:** Note the loss of the epithelium and extension of the ulcer down into the muscularis. There is normal gastric mucosa on the left then shelving down into a deep ulcer crater – see double ended arrow – whose base contains inflamed, necrotic debris.

![Microscopic Image](image2)

**Chronic gastritis:** the inflammatory cell infiltrates are composed mainly of lymphocytes and plasma cells, and occasionally some neutrophils. Mucosal atrophy and intestinal metaplasia are sequelae.

![Chronic Gastritis Image](image3)
**Helicobacter pylori:** this is a small, spiral, rod-shaped, gram negative bacterium residing under microaerobic conditions in a neutral microenvironment between the mucus and the superficial columnar mucosal cells. In the images below these bacteria are indicated by a short arrow. The organisms do not invade or directly damage the mucosa. These change the microenvironment of the stomach to promote mucosal damage. *H. pylori* contain urease and produce a protective surrounding cloud of ammonia to resist attack by gastric acid. The organisms are found in the surface epithelial mucus of most patients with active gastritis.

**Biochemistry:** nil

**Diagnosis:** barium meal, endoscopy with biopsy to exclude malignant change in the ulcer.

**Treatment:** antibiotics, sometimes for several weeks.

**Complications:** Patient may still need surgery if the ulcer is chronic, haemorrhaging, obstructing or perforating.

**Prognosis:** with treatment most patients get better.

**Trauma**

Trauma may be blunt or penetrating.

**Mechanism:** Blunt –
Midline Blunt Trauma – results typically in a combination of left hepatic lobe injury, pancreatic, proximal small bowel and mesentery injury, as well as central renal pedicle injury. If high forces are involved all viscera are damaged, including the aorta.

Right sided Blunt Trauma – results in right liver, right kidney, distal small bowel and mesentery, and right iliac wing injuries. There may also be a right sided haemopneumothorax.

Left sided Blunt Trauma – results in splenic and left renal injuries, injuries to the proximal small bowel and mesentery, the stomach and left iliac wing fractures and may cause a left sided haemopneumothorax.

Penetrating injuries: due to gun or knife wounds, industrial machine accidents and road traffic accidents.

The pattern of injury will depend on the entrance site, length and direction of the indwelling force.

Treatment nearly always requires surgery.

Causes of Oesophageal Trauma: commonest is instrumentation during endoscopy or the passage of nasogastric tubes.

Causes of gastric trauma: rare to be an isolated abdominal injury. Commonest cause is road traffic accidents. Occurred in 0.5% of cases of blunt abdominal trauma.

The majority of patients with proved bowel perforations do not have free gas on CT images. This occurs because the perforation seals spontaneously, because developing ileus prevents passage of gas into the abdominal cavity, or because small gas collections may rapidly be reabsorbed through the peritoneal lining.

Mortality is due to associated injuries.

Diaphragm tears: when on the left side the stomach can herniate through the defect.

In the CT images below – courtesy of JA Soto and SW Anderson, RSNA Radiology 265 (3) Dec 2012, the axial image shows the herniation of the stomach into the thoracic cavity. The stomach is pushed against the posterior ribs – called the ‘dependent viscera sign’ – see arrow. The sagittal reconstruction shows the defect in the diaphragm has created a waistlike constriction of herniated stomach – see arrow. This is called the ‘collar sign’.
Acquired Conditions

Neoplastic

Benign:

Polyps –

Definition: any mass that projects above the level of the surrounding mucosa.

Histology: 90% in the stomach are non-neoplastic and 10% are true adenomas. The latter have dysplastic epithelium and have malignant potential. Endoscopy with biopsy allows histological examination.

Malignant -

Gastric adenocarcinoma

Types: Adenocarcinomas comprise 90% of the malignant tumours of the stomach.

[The remainder are lymphomas 4%, carcinoid 3% and malignant stromal cell tumours (from the pacemaker cells of Cajal) 2%.

Site: 50% occur in the pylorus and 25% are on the lesser curve.

Incidence: 15th most common cancer in females and 11th most common cancer in Males.

Peak age: 60 years

Gender: M : F = 2 : 1.
Clinical Presentation: Early on, stomach cancer may cause indigestion, feeling bloated after eating a meal, heartburn, nausea and loss of appetite.

As the tumor grows, the patient may have stomach pain, blood in the stool, vomiting, weight loss, changed bowel habit, fatigue and swelling of the abdomen. If metastases have occurred to the liver, the patient may be jaundiced.

Molecular pathogenesis: genetic abnormalities in the intestinal type of gastric cancer include \( p53 \) mutation, abnormal E-cadherin expression and instability of \( TGF\beta \) and \( BAX \) genes.

Risk factors:

- Atrophic gastritis, especially patients with pernicious anaemia and the Plummer-Vinson Syndrome.
- Carcinogenic foods such as smoked foods, preserved meats and pickled foods
- Chronic gastritis of long standing can cause carcinoma. Transformation of benign chronic ulcers accounts for 10% gastric cancer.
- Genetic – said to be more prevalent in blood group A or AB.
- Benign tumours e.g. papillomas, adenomas if these are greater than 1.5 cm in diameter
- Geographic/ethnic factors: - high incidence in Austria, Chile, Finland, Iceland and Japan. Low incidence in Australia, UK and USA. Thought this is related to dietary factors.

Macroscopic appearance: 3 types – polypoid, ulcerating (70%), infiltrating
Microscopic appearance: 2 types – a) intestinal type which is common in atrophic gastritis.


Neoplastic glands infiltrate into the submucosa and some of the cells show mitoses – see arrow.

b) diffuse type which is poorly differentiated and produces the so-called “leather bottle stomach” – linitis plastica. This type has cells which secrete mucus, delivered to
the interstitium giving large pools of mucus (empty spaces). If mucus remains inside the cell, it pushes the nucleus to the periphery which has caused these to be called *signet-ring cells* – see arrows on 2 cells.

Image courtesy of WebPathol. Internet Pathology Laboratory for Medical Education, Mercer University School of Medicine 2015.

![Image](image_url)

**X-ray appearance:**

A barium meal can define at least 75% of gastric carcinomas.

Image below courtesy of Dr Martin Paz, Radiopaedia.org, rID 14183 shows cancer causing narrowing of the region of the pyloric antrum – see black arrows. Also the barium is streaking around the remnants of food in the dilated proximal stomach – white block arrow.

![Image](image_url)
An example below of the most common (70%) ulcerating type is courtesy of Dr Matt Skalski, Radiopaedia.org, rID21214. The barium has defined the irregular border to the antrum – see arrows and the portions that protrude into the lumen are irregular and considered ulcerating.

The polypoid type is demonstrated in the CT below, courtesy of Dr F Gaillard. Radiopaedia.org, rID 15047. The arrows indicate the border of the tumour which is projecting into the lumen of the stomach.

The infiltrating type – linitis plastica – in the CT below, courtesy of Dr David Cuete. Radiopaedia.org, rID 28080, is shown to be thickening the stomach wall and small projections extend to the left as early infiltration on that side – short white arrow. More extensive local extension is shown on the right of the stomach – long white arrow and there are multiple metastases in the liver – 2 black arrows.
Spread: occurs to involve the mucosa and stomach wall, fast invading into the serosa. Abdominal lymph nodes are involved early and also the cervical and mediastinal nodes. Once past the serosa, transcoelomic spread occurs with seeds lodging in the omentum, peritoneal wall and abdominal and pelvic organs. In females the ovaries are commonly involved where the metastases are called Krukenberg tumours. Blood spread can also occur early, especially to the liver, lungs, and bones.

Spread of Linitis plastica: in this situation, there is diffuse infiltration of the stomach by tumour. Metastases from breast carcinoma and lung carcinoma can behave the same as gastric carcinoma causing linitis plastica.

Biochemistry: examination of gastric cancer specimens using a combination of histopathology and magnetic resonance spectroscopy has identified glycine, alanine, free choline, and triglycerides as possible molecular markers related to the human gastric mucosa differentiation toward preneoplastic and neoplastic conditions. Ultrastructural studies of autoimmune atrophic gastritis and gastric adenocarcinoma revealed lipid accumulations intracellularly and extracellularly associated with a severe preneecrotic hypoxia and mitochondria degeneration.

Reference:

Diagnosis:
- gastroscopy and biopsy is the definitive method.
- Barium meal.
- CT scan shows spread into adjacent tissues and lymph nodes. If the wall of the stomach is greater than one cm and has a focal, eccentric abnormality which enhances this favours malignancy.
A full blood count may show anaemia if there has been bleeding from the malignancy and any bleeding may be detected by the faecal occult blood test.

**Treatment:** surgery, chemotherapy and sometimes radiotherapy.

**Prognosis:** gastric carcinoma now accounts for only 3% of cancer deaths whereas previously it was the leading cause of cancer death worldwide.

5-year survival of surgically treated early gastric carcinoma is 90% but only 15% for advanced cancer.

**Other malignant tumours of the stomach**

**Gastric lymphoma**

Accounts for 4% of stomach malignancies & 20% of extranodal lymphomas.

Gastric lymphomas are low grade B-cell neoplasms of the MALToma type and arise in a setting of chronic H.pylori gastritis with lymphoid hyperplasia. **This tumour can regress if H.pylori is treated.**

**B cell lymphoma**

**Barium Meal:** mass with narrowing of the lumen of the gastric antrum and a deep ulcer on the inferior wall (arrow). Arrowhead shows more nodules near the mass.

**Abdominal CT Scan.**

Arrowheads show the wall of the stomach to be thickened, with a lobulated inner surface and a smooth well defined outer wall.
Microscopy:

Blast cell infiltrate (short arrow) extending around glands (arrowhead). Mitoses – long arrow.

Carcinoid tumour

Carcinoid Tumour of the stomach – can secrete serotonin and metastasize causing the carcinoid syndrome.

Accounts for 1% of gastric neoplasms.

Type 1 – gastrin dependent, multiple, small, associated with chronic gastritis. No symptoms so often an incidental finding. 98% have 5 year survival.

Type 2 – gastrin dependent. Develop when there are gastrin secreting tumours e.g. gastrinoma of Zollinger-Ellison associated with MEN1. Have increased gastric acid, metastases to lymph nodes in 30%, liver metastases 10%. 90% 5 yr survival. Remove tumour.

Type 3 – gastric carcinoid (20%). Don’t have increased gastrin. Occur over the age of 50 years. Lymph nodes enlarged in 50%. 50% 5 yr survival but only 10% if have metastases.
Type 4 - gastric carcinoids – poorly differentiated endocrine carcinomas and mixed exocrine/endocrine carcinoma. Find atrophic gastritis in 50%. Size > 5 cm, ulcerating and unable to be resected. Poor prognosis – 8 months.

Gastrointestinal stromal tumours - GSIT

GIST is the most common mesenchymal tumour of the gastro-intestinal tract. They account for 5% of all sarcomas.

Includes leiomyoma, leiomyosarcoma, schwannomas, glomus tumours – all rare.

GISTs are believed to arise from the interstitial cells of Cajal, with 95% staining positive for CD117 (c-KIT) and 70% for CD34. The former is a tyrosine kinase growth factor receptor and the target of ST-571 (Imatinib; Glivec). It is thought these arise from the muscularis propria.

**Sites:** Ref: [http://www.histopathology-india.net/SoftTissuePathology.htm](http://www.histopathology-india.net/SoftTissuePathology.htm) Dr Sampurna Roy 2015

Presents as a primary tumour or metastatic mass. 25% of GISTs are malignant.

May be associated with von Recklinghausen's disease and Carney's triad (epithelioid gastric stromal tumour, extradrenal paraganglioma and pulmonary chordoma).

*Esophagus* - 5%
*Stomach* - 50-70%
*Small intestine* - 25-40%
*Colorectal* - Less than 10%
*Omentum, peritoneum, retroperitoneum* - about 7%

**Incidence:** 1.5 per 100,000 per year

**Mean age:** 60-65 years. Occurrence in children is very rare.

**Gender:** in children there is a female preponderance.

**Genetic:** families with germ-line autosomal dominant mutations of KIT or PDGFRA have been described presenting with multiple GIST at an early age.

**Clinical Presentation:** 69% detected due to symptoms, 21% incidental findings at surgery and 10% found at autopsy.

**Macroscopic appearance:**

Single or multiple tumours.

Size vary between 1 to more than 20cm.

Submucosal, intramuscular or subserosal mass.

Usually well circumscribed but lack a true capsule.

Cut surface grey to pink in colour.

Areas of cystic degeneration, infarction and hemorrhage and necrosis.

Prone to surface ulceration and bleeding.

An hourglass defect may occur at the pylorus or cardia if the tumour encircles the stomach.
**X-ray appearance:** These can be seen on barium studies with a large mass projecting into the lumen of the stomach as per the image below – courtesy of Sudhagar, Radiopaedia.org, rID 3893. Arrows indicate the perimeter of the bulky tumour within the gastric lumen.

![X-ray Image](image-url)

**Micro:** Have either the spindle-cell type or the epithelioid type.

**Spindle cells:**
Short. uniform, blunt ended, eosinophilic cytoplasm. Paranuclear vacuole may be present. Arranged in sheets, fascicles whorled, storiform or palisaded patterns.
Most gastric GISTs (corpus) and are spindle cell type.
The image below is courtesy of Dr F Gaillard. Radiopaedia.org, rID 36402

![Microscopic Image](image-url)

**Epithelioid cells:**
Epithelioid GISTs were previously known as epithelioid leiomyoblastoma.
Usually noted in the gastric antrum. Cells have abundant cytoplasm, round nuclei, cytoplasmic vacuoles and are arranged in sheets or nests. Clear cell, signet ring, oncocytic and plasmacytoid variants may be noted.

**Immunohistochemistry:**

Nearly always positive:
- CD34
- c-kit gene product (CD117) - confirms the diagnosis.
- Vimentin

Sometimes positive:
- Cytokeratin
- Smooth muscle actin and desmin
- S100 protein (especially in small bowel tumour)
- PGP9.5, neuron specific enolase, synaptophysin.

**Diagnosis:** relies on the morphology and the immunohistochemistry.

**Spread:** Staging procedures are based on the fact that most relapses affect the peritoneum and the liver.

Contrast-enhanced abdominal and pelvic CT scans are used to stage the tumour and also used for follow-up.

MRI provides better pre-operative stages for rectal GISTS.

FDG uptake in PET scans or FDG-PET-CT/MRI is mainly used to detect early response of the tumour to molecular targeted therapy.


**Treatment:** because of the rarity, multidisciplinary treatment planning is needed in centres handling a high number of patients annually - involving pathologists, radiologists, surgeons and medical oncologists.

*Localised disease:* complete surgical excision, without the dissection of clinically negative lymph nodes. A laparoscopic approach is not appropriate in patients who have large tumours, because of the risk of tumour rupture, which is associated with a very high risk of relapse. The aim is to have the excision margin without tumour cells.

This is followed by adjuvant treatment with imatinib (ST-571) for 3 years in patients who are high risk for relapse.

*Metastatic disease:* Imatinib is the standard treatment and continued indefinitely, since treatment interruption is followed by rapid tumour progression in nearly all cases, even when lesions have been previously surgically excised.
Prognosis: prognostic factors are the mitotic rate, tumour size and tumour site. Gastric GISTs have a better prognosis than those in small bowel or rectum. Tumour rupture represents a highly adverse prognostic factor as it will cause peritoneal seeding.

Based on people diagnosed between 2003 and 2009 the overall relative 5-year survival rate of people diagnosed with a malignant GIST was estimated to be about 76%.

- If the tumor was confined to the organ where it started, the 5-year relative survival was 91%.
- If it had grown into nearby tissue (or spread to nearby lymph nodes) when it was first diagnosed, the 5-year relative survival was around 74%.
- If it had spread to distant sites when it was first diagnosed, the 5-year relative survival was 48%.

Ref: American Cancer Society 2014.