Chapter 1
Achalasia

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Name and Synonyms

Achalasia

Incidence/Epidemiology

- Achalasia is a rare disease with an annual incidence of 1–2/100,000 persons.
- It affects men and women with equal frequency.
- It usually presents between the ages of 20 and 40, but may occur at any adult age.
- Onset before adolescence is rare.

Differential Diagnosis

- Achalasia is the most common motility disorder of the esophagus.
- It most often causes dysphagia (difficulty swallowing, as opposed to odynophagia—painful swallowing), so the differential is the differential for dysphagia.

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- Other diseases that may present with dysphagia and associated esophageal dilatation (secondary achalasia) include gastric cancer; infiltrative diseases such as amyloidosis, sarcoidosis, eosinophilic gastritis, and neurofibromatosis; and infections such as Chagas disease in South America.

Pathophysiology and Etiology

- Achalasia is a motility disorder of the esophagus; the cause is unknown.
- Histopathologically, achalasia is associated with degeneration of neurons in the myenteric plexus of the esophageal wall near the lower esophageal sphincter (LES). There is also an associated inflammatory response with an influx of inflammatory cells.
- The degeneration is most marked within the inhibitory neurons that allow for relaxation of the LES. This leads to a sustained contraction/increase in tone of the LES, resulting in an increase in LES pressure.
- Degeneration of the inhibitory neurons within the myenteric plexus in the lower two thirds of the esophagus affects the smooth muscle and causes aperistalsis.
- The cause of the degeneration is unknown. It has been ascribed to autoimmune phenomenon and sometimes to chronic viral infections from herpes simplex virus, measles virus, and HSV-1, but there is no conclusive evidence for any of these putative mechanisms.

Presentation

Typical/"Classic"

- Dysphagia (difficulty with swallowing) is the most common presenting symptom of achalasia, occurring in about 90 % of patients. Typically, the dysphagia is to solids and liquids equally.
- Most patients (about 75 %) also report regurgitation of undigested food. The regurgitated food remains in the esophagus, so usually there is no "acid taste" as in reflux. This has been described as a "bland reflux."
- Many patients describe an epigastric or retrosternal fullness after eating that may be severe enough to provoke attempts to induce vomiting, leading to confusion with eating disorders.
- Other commonly reported symptoms include difficulty belching, substernal chest pain associated with reflux, and frequent hiccups.
- Symptoms usually progress gradually. The mean time from symptom onset until diagnosis is about 5 years. This reflects the insidious nature of the symptoms, more than any particular difficulty in diagnosis.

- Some patients are noted to perform mechanical maneuvers (raising the arms above the head, standing erect and hyperextending the back) to help clear the esophagus. These actions may be noticed more by family members.

Atypical

- In retrospect, most patients will have years of somewhat typical symptoms before presentation.
- Most patients will have been treated for gastroesophageal reflux (GERD) before consideration of the diagnosis.
- Patients may present with chest pain, especially with eating. This may have been considered esophageal spasm and not responded to treatment.

Primary Differential Considerations

- Other causes of dysphagia should be considered, including
  - Esophageal tumors
  - Reflux /GERD
  - Connective tissue disorders, such as systemic sclerosis
  - Esophageal perforation
  - Esophageal spasm or stricture
  - Gastric cancer that involves the lower esophagus
  - Much rarer considerations:
    - Chagas disease
    - Plummer–Vinson syndrome

History and Physical Exam

Findings That Confirm Diagnosis

- The history usually is very suggestive of the diagnosis, although there are no absolutely confirmatory findings on history and physical exam. Physical exam usually is normal, although patients may have unintended weight loss.
- Confirmation is with esophageal manometry.

Factors That Suggest Diagnosis

- The historical features as described above suggest the diagnosis.
- An especially suggestive historical feature is dysphagia to solids and liquids equally.

Factors That Exclude Diagnosis

- Early in the course of illness, esophageal manometry may be nondiagnostic, so a high index of suspicion needs to be maintained if the symptoms continue or progress.

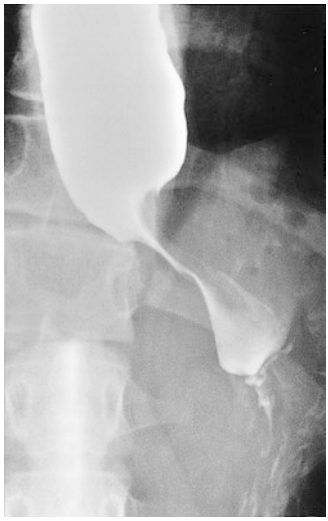
Ancillary Studies

Laboratory

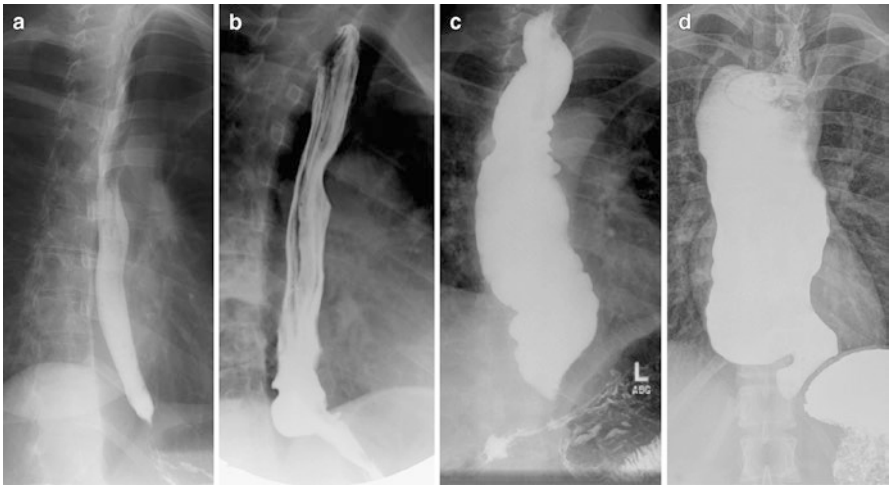
- Lab tests generally are not helpful in the diagnosis.

Imaging

- Chest x-ray may demonstrate widening of the esophagus or mediastinum, but more commonly is normal, especially early in the disease course.
- Barium swallowing studies may be helpful in suggesting the diagnosis, especially later in the course of illness.
- Classically, the barium esophagram demonstrates a proximally dilated esophagus that narrows to a contracted gastroesophageal junction, giving the classic "bird-beak" appearance of the esophagus.

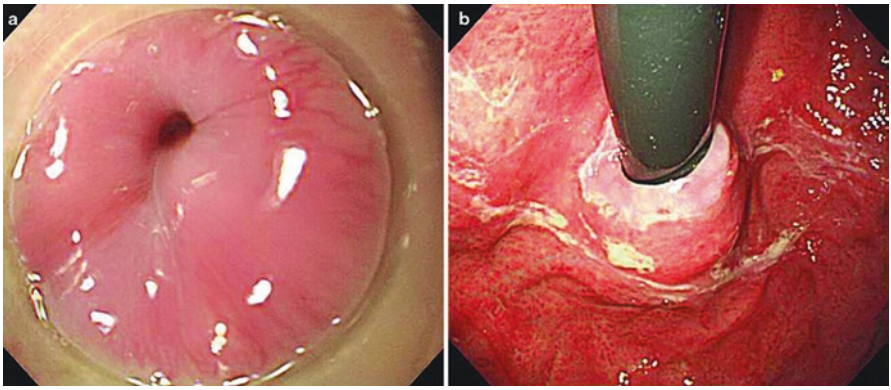


Classic "bird-beak" appearance of the esophagus on barium esophagram in achalasia. [Mittal R. Esophageal motor disorders. In: Orlando RC, editor. Atlas of esophageal diseases. 2nd ed. Philadelphia: Current Medicine; 2002. p 163–78.]



Examples of progressive dilation of the esophagus in different patients with achalasia. a Normal diameter esophagus leading to a bird's beak at the LES. b Minimal esophageal dilation (from 4 to 7 cm). c Progressive esophageal dilation (from 7 to 10 cm) with preserved esophageal axis. d Greater dilation (>10 cm) and initial sigmoidal course of the distal esophagus. [From article: A controversy that has been tough to swallow: is the treatment of achalasia now digested? J Gastrointest Surg. 2010 Feb;14 Suppl 1:S33-45. https://doi.org/10.1007/s11605-009-1013-5, at http://link.springer.com/article/10.1007%2Fs11605-009-1013-5; by Garrett R. Roll, Charlotte Rabl, Ruxandra Ciovica, Sofia Peeva, Guilherme M. Campos, © The Author(s) 2009; licensed under Creative Commons Attribution Noncommercial License https://creativecommons.org/licenses/by-nc/2.0/] *Caption from original*

- Other suggestive findings on barium study include a dilated esophagus, aperistalsis of the esophagus, and poor/delayed emptying of the barium into the stomach.
- Barium studies may be falsely negative in up to one third of cases.
- The diagnosis usually is confirmed by esophageal manometry, which is usually performed at specialized, experienced centers.
- Currently, the procedure of choice is high-resolution manometry (HRM) with or without esophageal pressure topography (HRMEPT).
- The classic finding on manometry is aperistalsis of the distal two thirds of the esophagus with incomplete relaxation of the LES. Increased pressure within the LES is a supportive finding but is not diagnostic.
- HRMEPT allows subtyping of achalasia, which may help in deciding treatment.
- Endoscopy (usually done when considering esophagitis, peptic ulcer disease, or gastritis in the diagnosis) usually is normal, though itmay demonstrate a dilated esophagus with retained products.



Endoscopic appearance of achalasia. (**a**) Sustained contraction of the lower esophageal sphincter as viewed from the proximal esophagus. (**b**) Retroflexed view of the LES from the stomach. [Park JM. Miscellaneous esophageal diseases. In: Chun HJ, Yang S-K, Choi M-G, editors. Clinical gastrointestinal endoscopy: a comprehensive atlas. Heidelberg: Springer. 2014. p. 87–98.] *Caption adapted from original*

Special Populations

Age

- Achalasia usually presents between the ages of 20 and 40 but may occur at any adult age.
- May be seen in adolescence, but is rare.

Pitfalls in Diagnosis

Critical Steps Not to Miss

- Consideration of the diagnosis is the first step.
- It is critical to consider malignancy in the differential diagnosis.

Mimics

- Any disease process that presents predominately with dysphagia can mimic achalasia.

- It is important to consider malignancy and other mechanical causes of dysphagia.
- Gastric cancer close to the gastroesophageal junction is a mimic of achalasia that must be considered.
- Space-occupying lesions (such as cancer) usually present with dysphagia that is greater with solids than liquids, but as they enlarge, they may produce dysphagia to both.
- Another diagnosis that may be confused with achalasia when evaluating a patient for dysphagia is myasthenia gravis. Myasthenia usually produces both dysphagia and dysphonia (decrease in tone/quality of voice).

Time-Dependent Interventions

- There are no particular time-dependent interventions in the evaluation of achalasia other than consideration of and evaluation for malignancy.

Overall Principles of Treatment

- Treatment is aimed at reducing the resting pressure of the LES.
- The first step usually is progressive, endoscopy-guided dilatation of the LES.
- If symptoms persist after three attempts at dilatation, or patients cannot tolerate dilatation, surgical myotomy often is performed.
- Patients who are not surgical candidates, or those who decline surgery, may be offered a trial of endoscopically guided direct injection of botulinum toxin into the LES.
- If all other treatments are unsuccessful, or the patient is not a candidate for surgery or endoscopy, a trial of oral nitrates or calcium channel blockers may be attempted.

Disease Course

- Achalasia is a chronically progressive disease; without treatment, esophageal dilatation will continue.
- Ten to fifteen percent of patients will develop end-stage disease even with treatment.
- Up to 5 % eventually may require complete esophagectomy, especially when the esophageal diameter exceeds 6 cm (megaesophagus).
- Patients with achalasia are at increased risk for squamous cell carcinoma of the esophagus. The absolute risk is low, however, so endoscopic surveillance is not recommended.

Related Evidence

Papers of particular interest have been highlighted as:
*** Of key importance*

Practice Guideline

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Cohort Study

Sadowski DC, Ackah F, Jiang B, Svenson LW. Achalasia: incidence, prevalence and survival. A population-based study. Neurogastroenterol Motil. 2010 Sep;22(9):e256–61. https://doi.org/10.1111/j.1365-2982.2010.01511.x. PMID: 20465592. http://www.ncbi.nlm.nih.gov/pubmed/20465592

Use PubMed Clinical Queries to find the most recent evidence. Use this search strategy:

"Esophageal Achalasia"[Mesh] OR "Achalasia"