

## Achalasia

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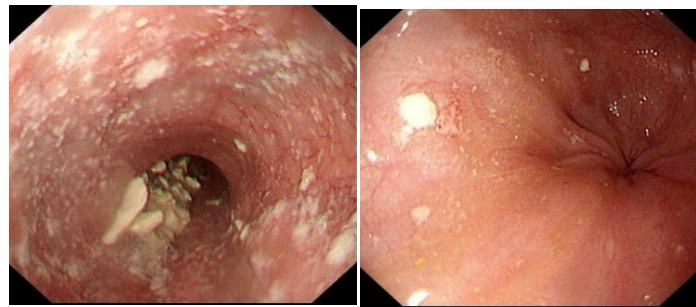
**Abstract:** A 53-year-old lady presents with worsening dysphagia to solids and liquids along with regurgitation. Endoscopy, barium swallow and manometry confirmed a diagnosis of achalasia, for which she successfully underwent a laparoscopic Heller myotomy and Dor fundoplication. The epidemiology, etiology, patho-physiology, investigation modalities, treatment options and follow-up of achalasia will be discussed.

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### 1. CASE REPORT

Mdm S is a 53-year-old chronic smoker of 30 pack-years with a past medical history of hypertension. She was referred by the polyclinic to the outpatient clinic complaining of a 2 year history of worsening dysphagia to both solids and liquids, which was associated with occasional regurgitation of undigested food. This was also accompanied with heartburn and weight loss of 10kg over the past 6 months. Her father passed away 30 years ago of lung cancer. Abdominal examination was unremarkable whilst chest x-ray (CXR) performed at the polyclinic did not show any widened mediastinum, air-fluid levels or pulmonary mass lesions.

Subsequently, she underwent an oesophago-gastro-duodenoscopy (OGD), which revealed a food and fluid-filled dilated esophagus and a tight lower esophageal sphincter (LES). There were no mass lesions and the scope was able to pass through easily (**Figure 1**). Barium swallow showed a narrowed gastro-esophageal junction (GEJ) with a classic “bird-beak” appearance (**Figure 2**). In view of her smoking and family history, computed tomography (CT) of the thorax was performed and it revealed a grossly dilated, fluid-filled esophagus to GEJ with no esophageal or pulmonary masses (**Figure 3**). High-resolution manometry confirmed esophageal aperistalsis with hypertensive LES which fails to relax adequately – findings consistent with type 1 achalasia.



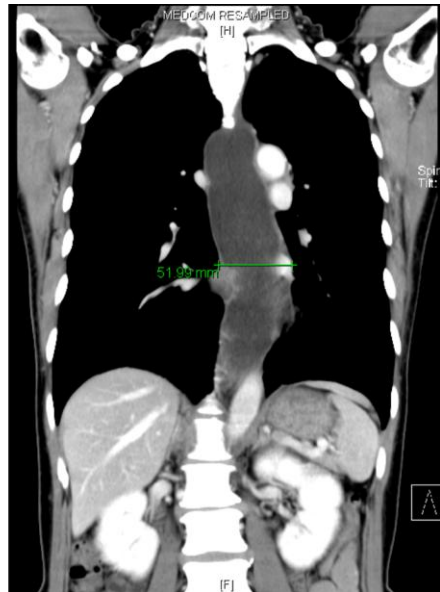
**Figure1.** Fluid-filled dilated esophagus (left) and a tight LES (right) on OGD



**Figure2.** Narrowed GEJ with a classic “bird-beak” appearance on barium swallow

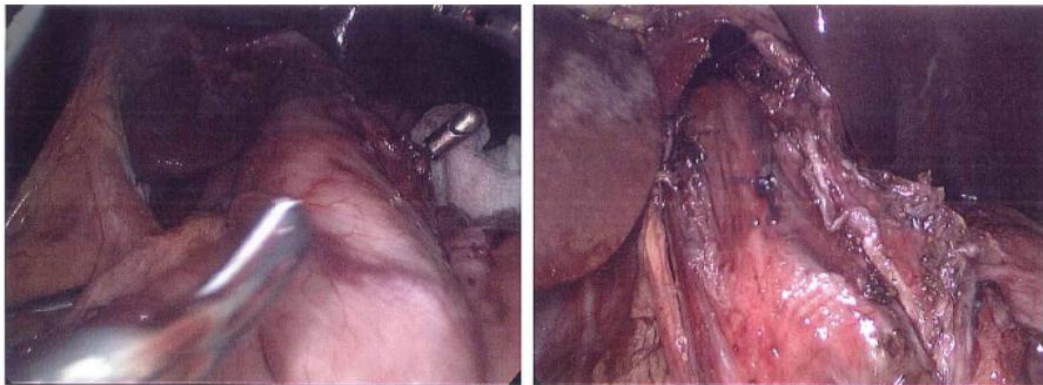
Mdm S was counseled and she consented for an elective laparoscopic Heller myotomy and Dor fundoplication. Intra-operatively, the proximal margin of constriction was identified with an on-table

OGD to be 35cm from the incisors, 6cm proximal to GEJ (**Figure 4**). 8cm of myotomy (adventitia and muscularis propria) was performed with Harmonics scalpel energy device from the proximal margin of constriction to 2cm distal to GEJ. The anterior and posterior branches of the vagus nerve were identified and preserved. A Dor fundoplication was then performed by plicating the gastric fundus anteriorly over the myotomy defect. Subsequent air-leak test was negative.



**Figure3.** Grossly dilated, fluid-filled esophagus on CT thorax

Post-operative recovery was uneventful. Barium swallow done on post-operative day (POD) 1 did not reveal any stricture or leak (**Figure 5**) and the patient was commenced on liquid diet. Mdm S was subsequently discharged well on post-operative day 2 and was well with nil further dysphagia at outpatient clinic follow-up.



**Figure4.** Dissection of GEJ (above left) with myotomy performed (above right) and Dor fundoplication (below left)



**Figure5.** POD 1 barium swallow with no stricture or leak

## 2. DISCUSSION

Esophageal achalasia is a primary esophageal motor disorder of unknown etiology. It is typified by failure of the LES to relax appropriately in response to swallowing with the decrease or absence of peristalsis [1]. Radiologically, it is characterized on a barium swallow by aperistalsis, esophageal dilation with minimal LES opening, “bird-beak” appearance and poor emptying of barium. Endoscopically, there is a dilated esophagus with retained saliva, liquid and undigested food particles in the absence of mucosal stricturing or tumor. Esophageal manometry will show incomplete or absent LES relaxation and the absence of esophageal peristalsis during swallowing and sometimes elevated resting LES pressure.

### *Epidemiology, Etiology and Patho-physiology*

Achalasia occurs equally in both men and women, with no racial predilection. Reported world-wide prevalence is 10 per 100,000 population [2], with Singapore’s prevalence at 1.8 per 100,000 population [3]. Regarding etiology, achalasia is often represented by the loss of the myenteric nerve fibers regulating inhibitory nitroergic neurotransmission in the LES, with the presence of a lymphocytic infiltrate and collagen deposition on histology [2]. The cause of this loss remains unclear. Proposed etiologies included that of infectious, hereditary and autoimmune causes. The only infectious agent identified as a cause of achalasia is *Trypanosoma cruzi*, responsible for Chagas’ disease. With regards to hereditary etiologies, achalasia has been reported to be part of a genetic syndromes such as Down syndrome, Allgrove syndrome and familial visceral neuropathy. Autoimmune causes of achalasia have been hypothesized to be associated with specific human leucocyte antigen (HLA) classes but no consistent association has been observed across studies. Despite increasing understanding of the physio- pathology of achalasia, its etiology remains largely unknown. Ultimately, the disease onset is characterized by chronic inflammation of the myenteric plexus of the esophagus secondary to an environmental insult. Genetic factors are probably involved in the development of achalasia, although the precise molecular basis of the disease has not been identified.

### *Investigation and Classification*

An assessment of esophageal motor function is essential in the diagnosis of achalasia. Barium swallow and OGD are complementary tests to manometry. The manometric techniques and equipment available in clinical practice ranges from conventional catheters with pressure sensors spaced anywhere from 3 to 5 cm apart or a water-perfused extrusion catheter to high-resolution manometry (HRM) assemblies that incorporate pressure sensors at 1 cm intervals [1]. The advent of HRM has allowed for the differentiation of achalasia into three subtypes, in accordance to the Chicago classification [4]:

- Type I (classic) – esophageal aperistalsis
- Type II – esophageal aperistalsis with intermittent compartmental pressurization
- Type III – esophageal spastic contractions

Types I and II probably represent a continuum of the natural history of the disease while Type III patients seem distinct from the others in that they are more akin to distal esophageal spasm, perhaps representing a variant of that disease that involves the LES [5].

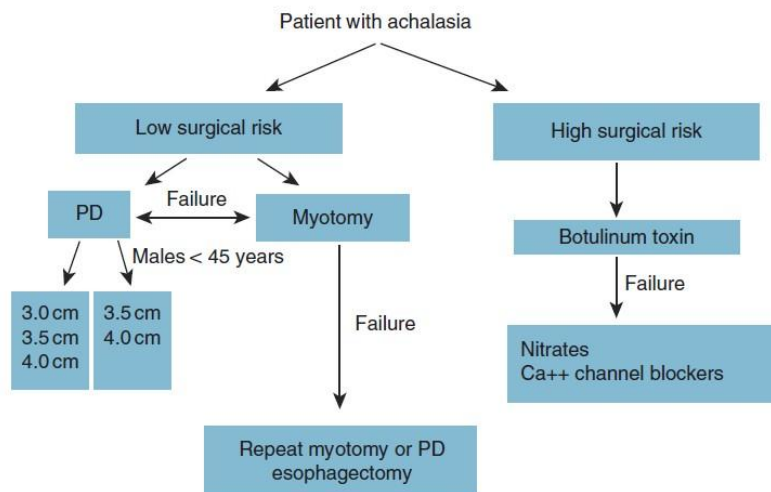
In a barium swallow, the diagnosis of achalasia is supported by findings of esophageal dilatation, GEJ narrowing with “bird-beak” appearance, aperistalsis, and poor contrast emptying. It is also useful to assess for late- or end-stage achalasia changes (tortuosity, angulation, mega-esophagus) that have implications for treatment. An additional role for radiological examination is to provide objective assessment of esophageal emptying after therapy. In many patients with treated achalasia, symptomatic relief does not always equate to functional esophageal emptying. Timed barium swallow, a measurement of barium column height 1 and 5 minutes after upright ingestion of a barium bolus, helps to identify patients who are more likely to have recurrence after treatment despite initial symptomatic improvement.

Endoscopy is primarily used to rule out a mechanical obstruction or pseudo- achalasia, which can mimic achalasia both clinically and manometrically. Endoscopic findings may range from a seemingly normal examination to a tortuous dilated sigmoid esophagus with retained food and saliva.

GEJ findings may also range from normal- appearing to a thickened muscular ring that may have a rosette configuration on retro- flexion view. Endoscopic ultrasound with histological confirmation should be performed in patients in whom there is a strong suspicion for malignancy.

### Treatment Options

Achalasia is a chronic condition without cure. Current treatments options are aimed at reducing the hypertonicity of the LES by pharmacologic, endoscopic or surgical means. The treatment goals are aimed to relieve patients' symptoms, improve esophageal emptying and prevent further dilation of the esophagus. **Figure 6** describes the recommended treatment algorithm for patients with achalasia [1].



**Figure6.** Achalasia treatment algorithm

PD: pneumatic dilatation

For patients who have low surgical risks, graded pneumatic dilation (PD) or laparoscopic surgical myotomy with a partial fundoplication are recommended as initial treatment options [1]. PD uses air pressures to intra-luminally dilate and disrupt the circular muscle fibres of the LES. The most common balloon dilator used for achalasia is the non-radiopaque graded size polyethylene balloons (Rigiflex dilators). The pressure required is usually 8 – 15 pounds per square inch (PSI) of air held for 15 – 60 seconds. The procedure is always performed under sedation and traditionally under fluoroscopy. Endoscopic balloon dilatation is often not employed because the largest endoscopic balloons employed have a diameter size of 2.0 cm, which limits their ability in causing LES disruption. Studies suggest that by using the graded dilator approach, good-to-excellent relief of symptoms is possible in 50 – 93 % of patients and cumulatively, dilation with 3.0, 3.5 and 4.0 cm balloon diameters results in good-to- excellent symptomatic relief in 74, 86, and 90 % of patients respectively, during an average follow-up of 1.6 years [1]. Esophageal perforation is the most serious complication associated with PD, with an overall median rate at 1.9 %. Conservative therapy with antibiotic, parenteral nutrition and stent placement may be sufficient for small perforations but surgical repair through a thoracotomy is the best approach in perforations with large and extensive mediastinal contamination.

First performed by Ernest Heller (1877 – 1964) in 1913, Heller myotomy involves division of the muscle fibres of the LES (circular layer) without disruption to the mucosa. With the progress of minimally-invasive surgery, this has resulted in decreased morbidity and faster recovery. Five different technical approaches have been described for the accomplishment of myotomy in achalasia patients: open transabdominal, open transthoracic, thoracoscopic, laparoscopic, and the robotic approach. There have been reports of good-to-excellent results of symptomatic relief in 60 – 94 % of patients who were followed for up till 36 years [1]. The development of gastro-esophageal reflux disease (GERD) after myotomy is a common post-operative side effect. Hence, whether a concurrent anti-reflux procedure should be performed has been extensively debated. The most recent achalasia guidelines from the Society of American Gastrointestinal and Endoscopic Surgeons (SAGES) recommended that patients who undergo myotomy should have a fundoplication to prevent reflux [6]. The most commonly used options for fundoplication after myotomy include an anterior Dor

fundoplication or a posterior Toupet fundoplication. With regards to surgery-related complications, esophageal perforation during surgery has been reported to occur on average in 6.9 % of patients but with clinical consequences in only 0.7 % of patients.

For patients who are not fit for any surgical intervention, endoscopic botulinum toxin (Botox) injection may be considered. Botox is a potent presynaptic inhibitor of acetylcholine release from nerve endings and this causes a short-term paralysis of the LES muscle. As it has no effect on the myogenic influence maintaining basal LES tone, treatment success is limited and approximately 50 % of patients relapse and require repeat treatments at 6 to 24-month intervals [1].

Although least effective, oral pharmacologic therapies are the last options available for patients who failed botox endoscopic injections. Calcium channel blockers (nifedipine) and long-acting nitrates (isosorbide dinitrate) are the two most common medications used to treat achalasia. They transiently reduce LES pressure by smooth muscle relaxation, facilitating esophageal emptying. The phosphodiesterase-5-inhibitor, sildenafil, has also been shown to lower the LES tone and residual pressure in patients with achalasia. Other less commonly used medications include anticholinergics (atropine, dicyclomine, cimetropium bromide),  $\beta$ -adrenergic agonists (terbutaline), and theophylline.

### **Follow-up**

Patient follow-up after therapy may include assessment of both symptomatic relief and esophageal emptying by barium swallow. On manometry surveillance, post treatment LES pressure of 10 mmHg or less is associated with a higher rate of remission [1]. The efficacy of both PD and Heller myotomy decreases over time. Repeat treatment will be required by some patients after 5 years. The subsequent choice of therapy may be best discussed through a multi-disciplinary team who can offer a multi- modality approach. In patient for which myotomy has failed, endoscopic botox injection may be effective. Up to 71% of such patients can have symptom resolution at 1 year post-injection [6]. On the other hand, PD is rarely used as salvage therapy following failed myotomy due to fears of increased perforation rates. Esophagectomy may be considered in appropriately selected patients after myotomy failure.

### **3. CONCLUSION**

The achalasia literature is limited due to the rarity of the disease. Patients who present with dysphagia to solids and liquids along with regurgitation must be investigated for achalasia. In a patient fit for surgery, the preferred treatment is surgical myotomy or pneumatic dilation. Long-term follow-up is required for surveillance of post- treatment efficacy, with LES pressure of 10 mmHg or less being the goal.

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