Achalasia Cardia Mimicking Oesophageal Cancer: A Revisit

Özofagus Kanserini Taklit Eden Akalazya Kardia: Tekrar Değerlendirme

Muhammad Nur Syamim Che Johan[1], Firdaus Hayati[1], Allim Khairuddin[2], Nornazirah Azizan[3]

[1]Department of Surgery, Faculty of Medicine and Health Sciences, Universiti Malaysia Sabah, Kota Kinabalu, Sabah, Malaysia
[2]Department of Surgery, Queen Elizabeth Hospital, Ministry of Health Malaysia, Kota Kinabalu, Sabah, Malaysia
[3]Department of Pathobiology and Medical Diagnostic, Faculty of Medicine and Health Sciences, Universiti Malaysia Sabah, Kota Kinabalu, Sabah, Malaysia

ABSTRACT

Achalasia results from a progressive degeneration of ganglion cells in the myenteric plexus of the oesophagus. Patients commonly presented with insidious onset of dysphagia and sometimes may mimic oesophageal cancer (pseudoachalasia). A 36-year-old lady presented with dysphagia of both liquid and solid for 3 months duration. Upper endoscopy is one of the diagnostic evaluations of achalasia but oesophageal manometry remains the gold standard in diagnosing achalasia. Resection (esophagectomy) gives better outcome in patients with grossly dilated distal esophageal as reported in this case.

Keywords: Oesophagectomy, Oesophageal Achalasia, Oesophageal Neoplasms

Received: 01.29.2021 **Accepted:** 06.04.2021

ÖZET

Akalazya, yemek borusunun myenterik pleksusunda ganglion hücrelerinin ilerleyici dejenerasyonundan kaynaklanır. Hastalar genellikle sinsi başlangıçlı disfaji ile başvururlar ve bazen özofagus kanserini (psödoakalazya) taklit edebilirler. 36 yaşında bayan hasta 3 aydır hem sıvı hem de katı disfaji şikayeti ile başvurdu. Üst endoskopi, akalazyanın tanısal değerlendirmelerinden biridir, ancak özofagus manometrisi, akalazya tanısında altın standart olmaya devam etmektedir. Rezeksiyon (özofajektomi), bu vakada bildirildiği gibi, büyük ölçüde dilate distal özofaguslu hastalarda daha iyi sonuç verir.

Anahtar Sözcükler: Özofajektomi, Özofagus Akalazya, Özofagus Neoplazmaları

Geliş Tarihi: 29.01.2021 **Kabul Tarihi:** 04.06.2021

ORCID IDs: S.J.0000-0002-9797-9352, F.H.0000-0002-3757-9744, N.A.0000-0002-5831-3734

Address for Correspondence / Yazışma Adresi: Muhammad Nur Syamim Che Johan, MD Department of Surgery, Faculty of Medicine and Health Sciences, Universiti Malaysia Sabah, Kota Kinabalu, Sabah, Malaysia E-mail: syamimjohan76376@gmail.com

doi:http://dx.doi.org/10.12996/gmj.2021.109

INTRODUCTION

Achalasia results from progressive degeneration of ganglion cells in the myenteric plexus of the oesophagus. It will lead to a failure of relaxation of the lower esophageal sphincter (LES), accompanied by a loss of peristalsis in the distal esophagus. It is an uncommon disorder with an annual incidence of approximately 1.6 cases per 100,000 individuals and the prevalence of 10 cases per 100,000 individuals (1). It can be subdivided into either primary or secondary achalasia. The aetiology of primary or idiopathic type is unknown. Secondary achalasia occurs due to diseases that cause esophageal motor abnormalities similar or identical to that primary achalasia. Most patients usually present with a similar clinical presentation; dysphagia for liquids first then solids and regurgitation of bland undigested food or saliva.

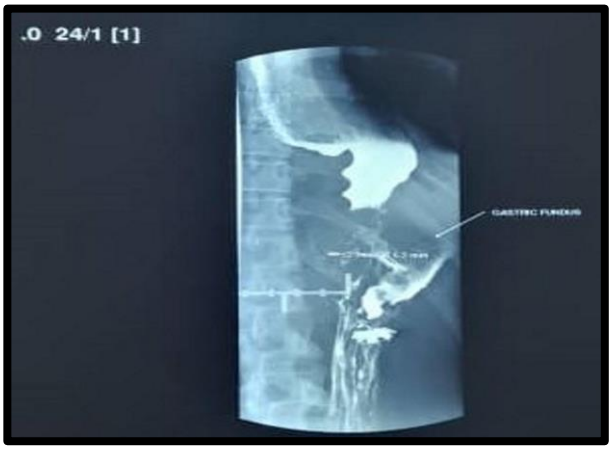
Diagnosis can be achieved by thorough history taking and physical examination in addition to imaging studies namely barium swallow and computed tomography (CT) scan and endoscopic evaluation. Esophageal manometry is required to establish the diagnosis, especially to rule out achalasia. However, certain differential diagnoses might not be easy by routine methods in the initial evaluation as the clinical presentations can mimic idiopathic type resulting in a substantial delay in diagnosis and treatment. Herein, we describe a middle-aged lady with classical presentations of oesophageal carcinoma but histology was consistent with achalasia cardia.

CASE REPORT

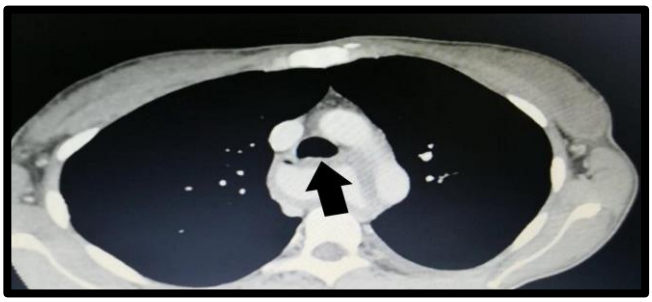
A 36-year-old lady with no known medical illness presented with a 3-month history of progressive dysphagia to both solids and liquids. It was associated with loss of weight and loss of appetite. She has no family history of malignancy. Upper endoscopy revealed esophagitis with cardia swelling (suspicious of achalasia) and gastritis. Random biopsy was taken over gastric, cardia and lower esophageal mucosa and showed *Helicobacter pylori* gastritis with no evidence of metaplasia and dysplasia. Repeated upper endoscopy was performed a month later showed gastroesophageal junction (GOJ) at 45 cm, 41 cm from incisor noted outpouching externally, unable to enter scope more than 10mm. Contrast injection revealed extravasation of contrast into a 15mm pocket adjacent to GOJ. Manometry study showed type I achalasia. Barium swallow was performed (Figure 1A) showing a short tight gastroesophageal stricture with proximal esophageal dilatation and tertiary contraction. However, in view of irregularity at the gastroesophageal region and gastric fundus, malignancy is a consideration. Further imaging was done which was contrast-enhanced CT abdomen (Figure 1B) showed grossly dilated esophagus causing hold up of the oral contrast. Food particles are present in the distal esophagus. The dilated esophagus also causes compression of the left lower lobe bronchus- lower lobe consolidation. Given the imaging features, they are suggestive of achalasia cardia.

We had performed a 3-staged McKeown esophagectomy. Intraoperatively, post thoracoscopic esophageal mobilization while proceeding with the abdominal part of gastric mobilization it was noted that there was a huge distal esophageal mass, hence causing the chronic dysphagia form external compression at the distal oesophagus rather than true achalasia. Decision made in conjunction with consent ie possibility of conversion to open, left thoracotomy was performed to have adequate access to the distal oesophagus and the tumour margins. 12 x 10 cm of tumor distal esophagus with thickened esophagus proximally (Figure 2A-2B). The tumour attached and closely adherent to the left diaphragm. Resection around its margin the diaphragm was left with a big defect; an approximation of the diaphragmatic muscle made just enough to allow the gastric tube passing through.

The macroscopic evaluation revealed no definite tumour identified upon the cut section. Instead, there was an area of the thickened esophageal wall. Histopathological examination showed diffuse muscular hypertrophy with distal aganglionosis consistent with underlying achalasia cardia (Figure 3A-3C). No dysplasia or malignancy was noted. The postoperative recovery was uneventful. She was discharged after 1 week following enhanced recovery after surgery protocol. Upon follow up at 4 weeks, she was well without any surgical complication or recurrence.

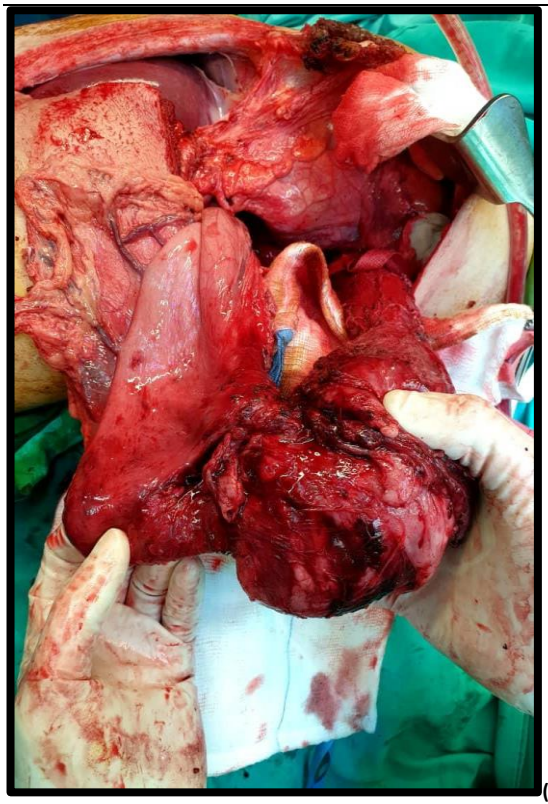


(A)

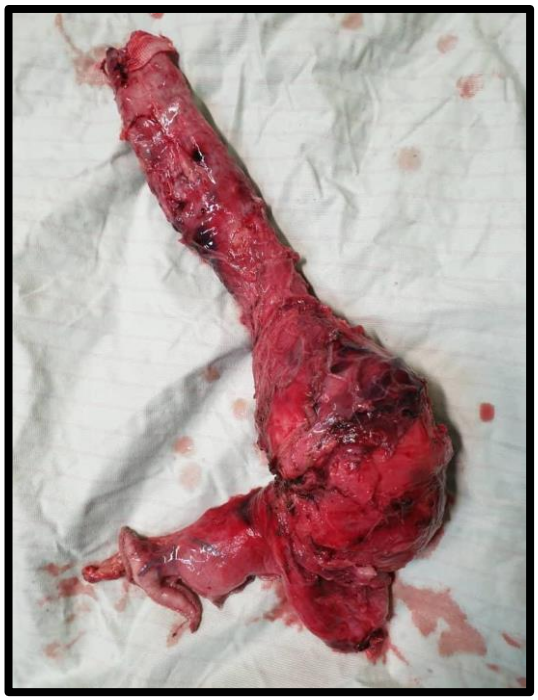


(B)

Figure 1: (A) Barium swallow showing a short tight gastroesophageal stricture with proximal esophageal dilatation and tertiary contraction. (B) Contrast enhanced CT thorax showing grossly dilated distal esophagus (arrow) in which causes compression of the left lower lobe bronchus.



(A)



(B)

Figure 2: (A) 12 x 10 cm tumor distal esophagus with thickened esophagus proximally (B) Gross specimen of total esophagectomy with tumor over distal esophagus.

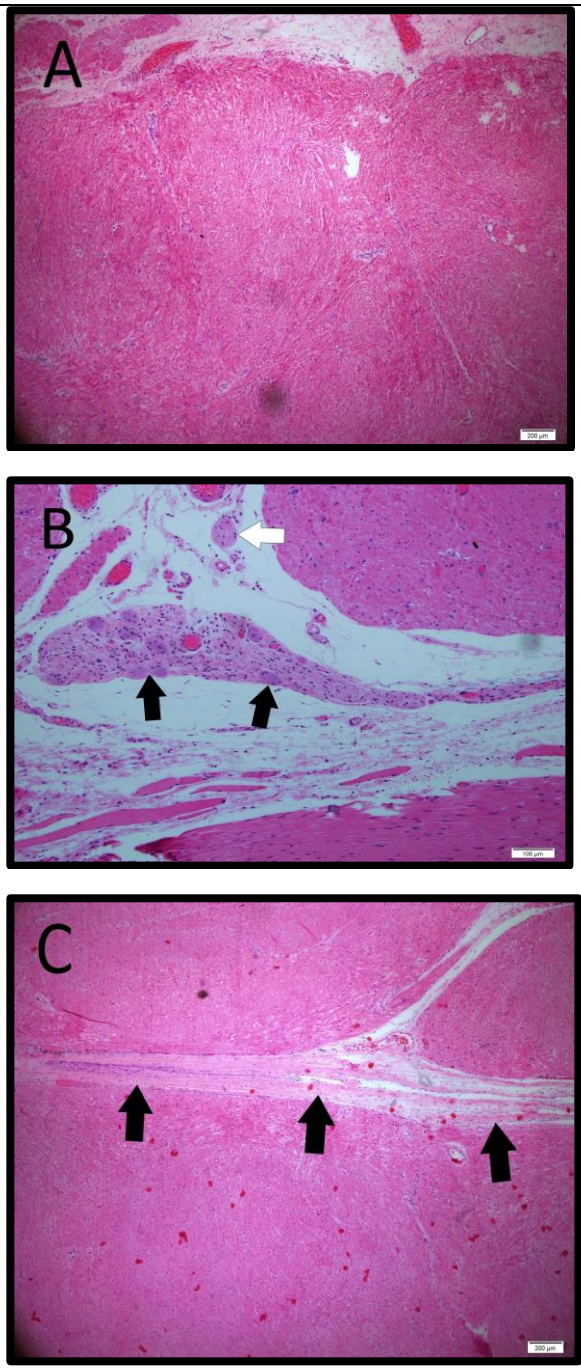


Figure 3: (A) Hypertrophic muscle bundle at gastro-oesophageal junction. (B) Presence of ganglion cells (black arrow) and nerve bundles (white arrow) at the proximal part of oesophagus. (C) Aganglionic distal oesophagus along the submucosa (black arrow).

DISCUSSION

Achalasia derives from a Greek word which means “failure of relaxation”, first coined by Hurst in 1927 (2). Both primary and secondary achalasia are clinically important however, another important similar condition that should not be neglected by the clinician is pseudoachalasia. It is a clinical syndrome due to malignant tumors mimicking an idiopathic type. Its pathophysiology is also not well understood, nevertheless few theories have been postulated. Intraluminal narrowing, extraluminal compression, infiltration of the myenteric plexus, metabolic disturbance, and paraneoplastic effects as endocrine abnormalities are among the predicted causes (3).

It is of utmost importance to rule out malignancy especially gastro-oesophageal junction adenocarcinoma, as the majority can present as benign intramural lesion without superficial macroscopic lesion (2). Other malignancies namely gastric cancer, bronchial adenocarcinoma, esophageal squamous cell carcinoma, Hodgkin / non-Hodgkin's lymphoma of the mediastinum, pleural mesothelioma, and mediastinal lymphadenopathy can be the differential diagnoses as they lead to extraluminal compression as well as infiltration to the myenteric plexus (2). In our case, all investigations performed directing towards a malignancy.

Achalasia has an insidious onset, and disease progression is gradual. Patients typically experience symptoms for years prior to seeking medical attention. In one series of 87 consecutive patients with newly diagnosed achalasia, the mean duration of symptoms was 4.7 years prior to the diagnosis (4). Progressive dysphagia from liquid to solid is the most characteristic symptom of primary achalasia. However, dysphagia in achalasia due to malignancy is relatively short in duration and usually develops in advanced phases of pathology. In our case, she complained of dysphagia of both liquid and solid, which is difficult to be determined as typical symptoms of achalasia. Although a secondary form of achalasia involving the cardia can be easily diagnosed through upper gastrointestinal endoscopy. But, if it spreads into the submucosal layer or is a very aggressive and poorly differentiated histological type, it can be very difficult to diagnose it until surgery.

Rapid progression of dysphagia and profound weight loss are suggestive of pseudoachalasia due to a malignancy. There are case reports noting a rare condition called pseudoachalasia. This is defined as signs and symptoms consistent with primary achalasia but which is due instead to the invasion of the distal oesophagus or its neural plexus by malignant tumours. Pseudoachalasia has been mostly associated with gastric adenocarcinomas or squamous cell carcinomas of the oesophagus but rarely as adenocarcinomas of the oesophagus.

Diagnostic evaluation of patients with suspected achalasia consists of upper endoscopy, esophageal manometry, barium swallow and EUS (endoscopic ultrasound). Our patient initially was reported to have oesophagitis with cardia swelling. No typical presentation of achalasia cardia were found, namely a retained food in the oesophagus or an unusually increased resistance to passage of endoscope through oesophagogastric junction. Hence, oesophageal manometry was performed showing type I achalasia. To date, oesophageal manometry remains the gold standard in diagnosing achalasia. (5). An imaging scan (contrasted CT) can help in diagnosing patients with suspected achalasia or pseudoachalasia. CT evaluation of achalasia has apparently been limited, with little information in the radiologic literature. However it is important to recognize the characteristic CT features of achalasia in order to distinguish it from other entities causing oesophageal symptoms (6).

General aim in managing patients with achalasia cardia is aimed at decreasing the resting pressure in the LES to a level at which the sphincter has no longer impedes the passage of ingested material. This includes mechanical disruption of the muscle fibres of LES or by pharmacological reduction in LES pressure. Studies comparing medical and endoscopic therapies to surgical management have demonstrated superior results with surgery (7,8). This case report presents an achalasia cardia patient with grossly dilated distal esophageal mimicking an esophageal tumor. Indication for resection in this case is grossly dilated distal esophagus as reported by CT imaging. Resection (oesophagectomy) was recommended for patients with more than one failed myotomy or a markedly dilated esophagus.

The finding of cancer of the oesophagus during the workup of recurrent dysphagia would obviously be an indication for resection as well as peptic stricture from reflux. Besides, larger reports have found the most frequent indications for esophagectomy in the setting of achalasia to be tortuous megaesophagus (64%), failure of prior myotomies (63%), and associated reflux strictures (7%) and favorable outcomes were observed following resection (9).

CONCLUSION

Achalasia, a primary oesophageal disease is frequently insidious with dysphagia, regurgitation or chest pain. A barium swallow may show pathognomonic features of achalasia but oesophageal manometry is a gold standard. Grossly dilated esophageal from long term of achalasia may be treated by surgical resection (esophagectomy) in which giving a better outcome compared to other treatment modalities.

Conflict of interest
No conflict of interest was declared by the authors.

Acknowledgement
We would like to thank the Director General of Health Malaysia for his permission to publish this article as a case report.

REFERENCES

1. Sadowski DC, Ackah F, Jiang B, Svenson LW. Achalasia: incidence, prevalence and survival. A population-based study. Neurogastroenterol Motil. 2010; 22: e256.
2. Agrusa A, Romano G, Frazzetta G, De Vita G, Chianetta D, Di Buono G, et al. Achalasia secondary to submucosal invasion by poorly differentiated adenocarcinoma of the cardia, Siewert II: Consideration on preoperative workup. Case Rep Surg. 2014; 2014: 654917.
3. Pardi DS, Miller SM, Miller DL, Burgart LJ, Szurszewski JH, Lennon VA, et al. Paraneoplastic dysmotility: loss of interstitial cells of Cajal. Am J Gastroenterol. 2002; 97(7): 1828-33.
4. Jeon HH, Kim JH, Youn YH, Park H, Conklin JL. Clinical characteristics of patients with untreated achalasia. J Neurogastroenterol Motil. 2017; 23(3): 378–384.
5. Pohl D, Tutuian R. Achalasia: an overview of diagnosis and treatment.J Gastrointestin Liver Dis. 2007; 16(3): 297-303.
6. Rabushka LS, Fishman EK, Kuhlman JE. CT evaluation of achalasia. J Comput Assist Tomogr. 1991; 15(3): 434-9.
7. Anselmino M, Perdikis G, Hinder RA, Polishuk PV, Wilson P, Terry JD, et al. Heller myotomy is superior to dilatation for the treatment of early achalasia. Arch Surg. 1997; 132(3): 233-40.
8. Malthaner R, Todd TR, Milner L, Ranson FG. Long-term results in surgically managed esophageal achalasia. Ann thoracic Surg. 1994; 58: 1343-1347
9. Devaney EJ, Orringer MB. Esophagectomy for achalasia: patient selection and clinical experience. Ann Thorac Surg. 2001; 72: 854-858.