Achalasia Cardia: A Case Report in Young Female

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Abstract

Introduction

Achalasia results from the degeneration of inhibitory ganglion cells within the esophageal myenteric plexus and the lower esophageal sphincter (LES), leading to a loss of inhibitory neurons and resulting in the absence of peristalsis with failure of LES relaxation. Its origins are multifactorial, potentially involving infections, autoimmune responses, and genetics, with equal incidence in males and females. The hallmark symptom

symptoms include progressive dysphagia for solids and liquids, along with regurgitation, heartburn, and non-cardiac chest pain.

Case Presentation

A 22-year-old female patient initially diagnosed with gastroesophageal reflux disease(GERD) received proton pump inhibitors and antacid gel for persistent dysphagia and regurgitation. Subsequent tests including barium esophagogram and manometry indicated Type II Achalasia Cardia. The patient showed clinical improvement with relief of dysphagia, regurgitation, and heartburn symptoms after pneumatic balloon dilatation (PBD). She was advised to follow up after six months with upper GI endoscopy and manometry in the outpatient clinic for regular endoscopic surveillance as there is a risk of transformation to esophageal carcinoma.

Case Discussion

Diagnosing achalasia in young adults poses challenges due to its diverse presentation and resemblance to other esophageal disorders like GERD. Diagnosis relies on clinical symptoms and imaging studies such as barium esophagogram revealing a bird's beak appearance and esophageal manometry showing absent peristalsis. Upper GI endoscopy is needed to rule out malignancy. Treatment options include non-surgical approaches like medication and Botox injections, as well as surgical methods such as pneumatic balloon dilation, laparoscopic Heller myotomy, and per-oral endoscopic myotomy (POEM). The treatment options depend upon the patient's condition at presentation and their individual choices.

Conclusion

This case report emphasizes that it is crucial to consider achalasia as a potential differential diagnosis in young adults with dysphagia, especially if conventional treatments for acid peptic disorder do not alleviate symptoms. Prompt diagnosis and appropriate management can lead to significant clinical improvement and better patient outcomes.

KEYWORDS

Achalasia, dysphagia, manometry, pneumatic dilatation, case report

Key Clinical Message

This case of a 22-year-old female emphasizes the significance of considering achalasia in young adults with persistent dysphagia despite conventional treatments for acid peptic disorder. Prompt diagnosis and pneumatic dilation intervention led to clinical improvement, highlighting the importance of early recognition and appropriate management for better patient outcomes.

INTRODUCTION

Achalasia, a rare motor neuron disorder, presents an annual incidence of 1 per 100,000 individuals and a prevalence of 10 per 100,000.[1] It is characterized by the degeneration of inhibitory ganglion cells within the esophageal myenteric plexus and the lower esophageal sphincter (LES), leading to a loss of inhibitory neurons and resulting in the absence of peristalsis with failure of LES relaxation .[2] Literature suggests a multifactorial origin of achalasia, implicating infectious agents, autoimmune responses, and genetic factors as potential triggers .[3] While achalasia affects males and females equally, slightly higher rates in females have been reported.[4,5] The hallmark symptom associated with achalasia is slowly progressive dysphagia for both solids and liquids, experienced by the majority of affected patients, accompanied by regurgitation, heartburn, and non-cardiac chest pain .[6,7]

Diagnosis of achalasia relies on peculiar clinical symptoms and imaging studies such as barium swallow and esophageal manometry .[8] A barium esophagogram reveals classic findings like the "bird's beak" appearance and esophageal dilation which serve as the primary diagnostic tool .[9] Esophageal manometry is the gold standard method demonstrating absent peristalsis . Upper endoscopy is further recommended to exclude

malignancy.[10]Treatment strategies encompass both nonsurgical options like pharmacotherapy (calcium channel blockers, nitrates, and botulinum toxin) and surgical techniques such as pneumatic dilation, laparoscopic Heller's myotomy, and per-oral endoscopic myotomy(POEM), aiming to alleviate symptoms while considering the risk of reflux.[11] We present a case involving a young patient initially misdiagnosed and treated for acid peptic disorder (APD), which was later diagnosed with achalasia cardia following comprehensive evaluation. Subsequent pneumatic dilation resulted in the alleviation of her symptoms. Due to its rarity, achalasia often presents a challenge for early diagnosis, leading to delayed recognition and commonly mistaken symptoms for other upper digestive issues such as gastroesophageal reflux .

CASE PRESENTATION

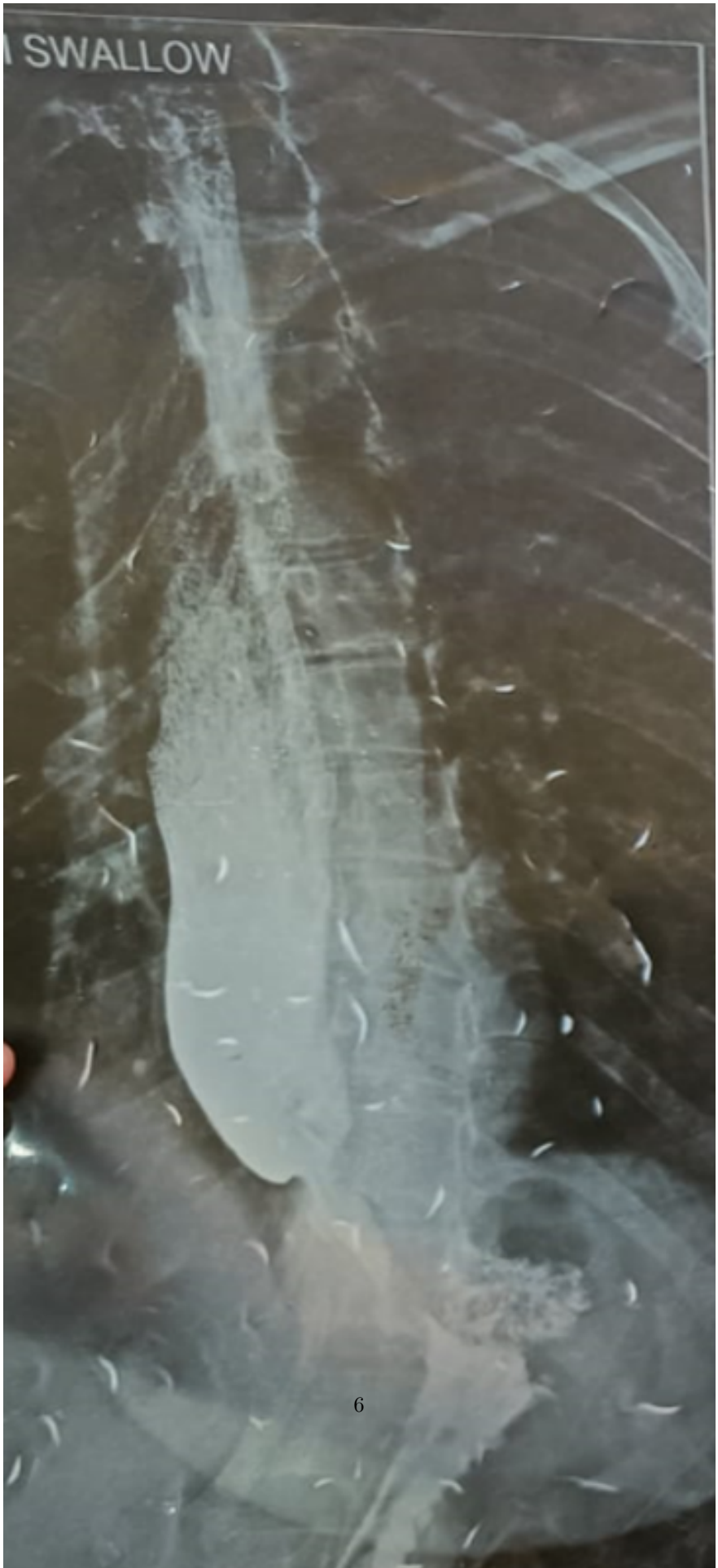
A 22-year-old female patient from Hetauda presented to OPD on 7th December 2018, with a one-month history of persistent dysphagia for both solids and liquids, accompanied by nausea and vomiting of undigested food. She also gave a history of weight loss of five kilograms in the last three months. She reported experiencing similar symptoms intermittently over the last two years and her condition did not improve with antacid treatment. The patient does not have any reported allergies and is not currently taking any medications. Importantly, there is no notable medical or family history, and psychosocial factors were considered irrelevant to the presentation. A thorough physical examination revealed normal vital signs, absence of palpable masses or tenderness upon abdominal assessment, and intact cranial nerve functions, as well as normal motor and sensory responses on neurological examination, highlighting the need for further diagnostic investigations.

Subsequent diagnostic assessments including upper gastrointestinal (UGI) endoscopy, barium esophagogram, and manometry, were planned to elucidate the underlying cause of the patient's distressing symptoms. Initially, an upper gastrointestinal endoscopy performed on December 8, 2018, detected dilation in the lower portion of the esophagus (Fig-3). On December 9, 2018, a barium esophagogram performed revealed narrowing at the distal thoracic gastroesophageal junction with dilation of the mid and distal thoracic esophagus, causing mild anterior displacement of the trachea and carina(Fig-1). Manometry confirmed ineffective esophageal motility showing raised Integrated Relaxation Pressure(IRP) with failed peristalsis in all swallows and pan esophageal pressurization suggestive of type II Achalasia Cardia(Fig-2). The patient was thoroughly explained about the diagnosis, the treatment options available, and potential complications if left untreated. She was prescribed Pantop 40 mg per oral once daily, Ondem 4mg as needed, and sucralfate 10ml per oral thrice a day for two months. Since advanced surgical techniques such as pneumatic balloon dilatation were not accessible at that facility, the patient was referred to a tertiary center in Kathmandu for additional treatment.

On June 6, 2019, about six months after diagnosis, the patient and her guardian sought care at a tertiary center in Kathmandu. During these six-month periods, she visited multiple healthcare facilities for her regurgitation and received antacid therapy for reflux. After assessing her symptoms and examining her reports, she was presented with options of pneumatic balloon dilatation (PBD) or surgical myotomy, accompanied by comprehensive discussions outlining the advantages and disadvantages of each procedure. She opted for PBD and was admitted on June 7, 2019, where she followed a clear liquid diet regimen before undergoing the procedure two days later

The PBD procedure was done on 9th June 2019 by sedating the patient with midazolam (15 mcg/kg, IV) and propofol (2 mg/kg, IV). Her vital signs were monitored throughout the procedure by the team of anesthesiologists and gastroenterologists performing it. Upper GI endoscopy revealed a dilated esophagus with a relatively tight lower esophageal sphincter (LES) but no obvious stricture. A foreign body of meat bolus was found and removed with a dormia basket. The second part of the Duodenum(D2), fundus, and pseudo-achalasia were ruled out. The esophagogastric junction (ECJ) was identified at 40 cm from the incisor and marked accordingly on the Rigiflex 30 mm balloon. A guidewire was placed across the ECJ, and the rigiflex balloon was introduced. The balloon was dilated with 16 psi until the disappearance of the waist for about a minute. At the end of the procedure, the balloon was withdrawn, revealing a tinge of fresh blood on it. Post-procedure gastroscopy was repeated revealing a relatively easily giving LES and no obvious tear or active bleeding (Fig 4). Following the procedure, the patient experienced notable clinical improvement, with

relief from dysphagia, and regurgitation during subsequent assessment. Regular follow-up appointments were scheduled to monitor the long-term efficacy of the intervention, assess adherence, and evaluate tolerability. Additionally, vigilance for potential adverse events such as esophageal perforation or bleeding was prioritized to ensure timely detection and management.



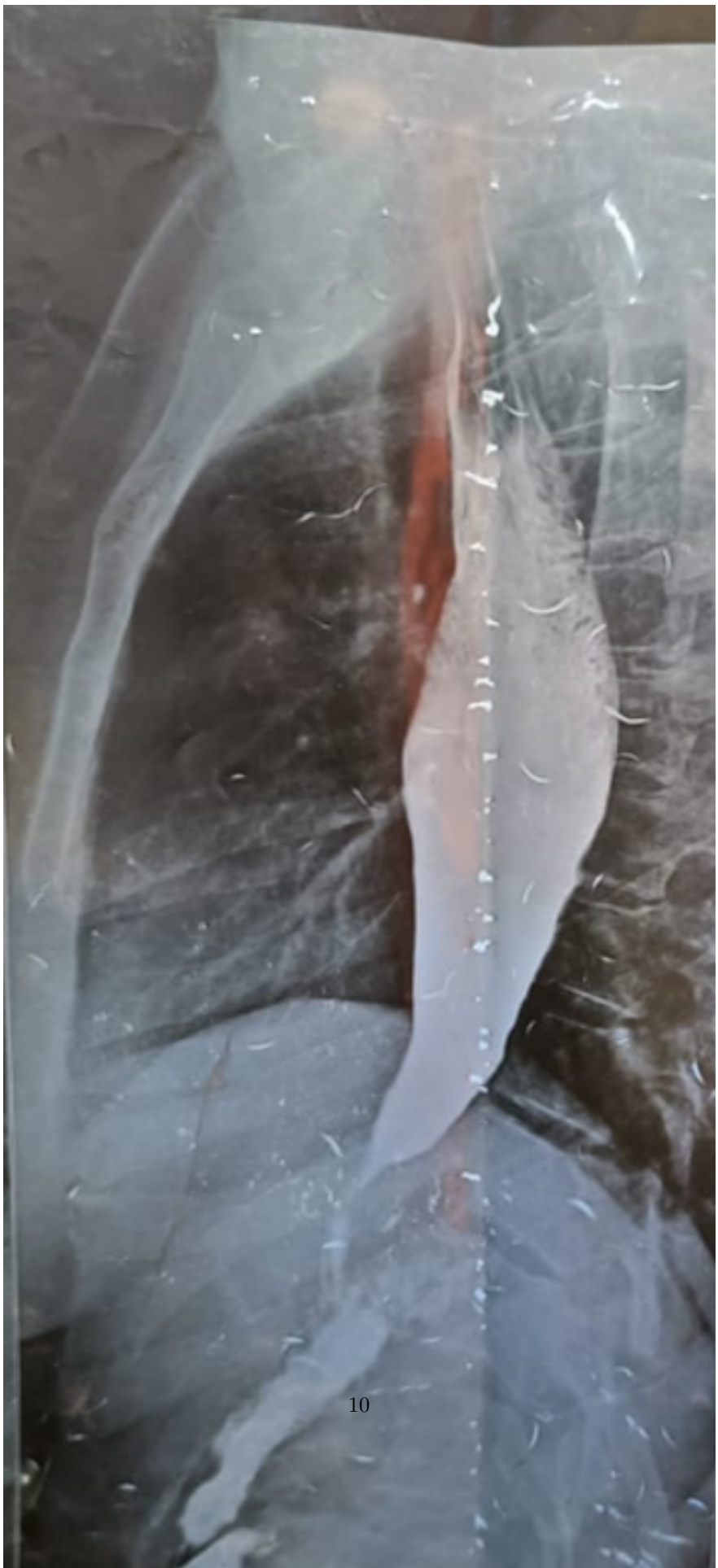
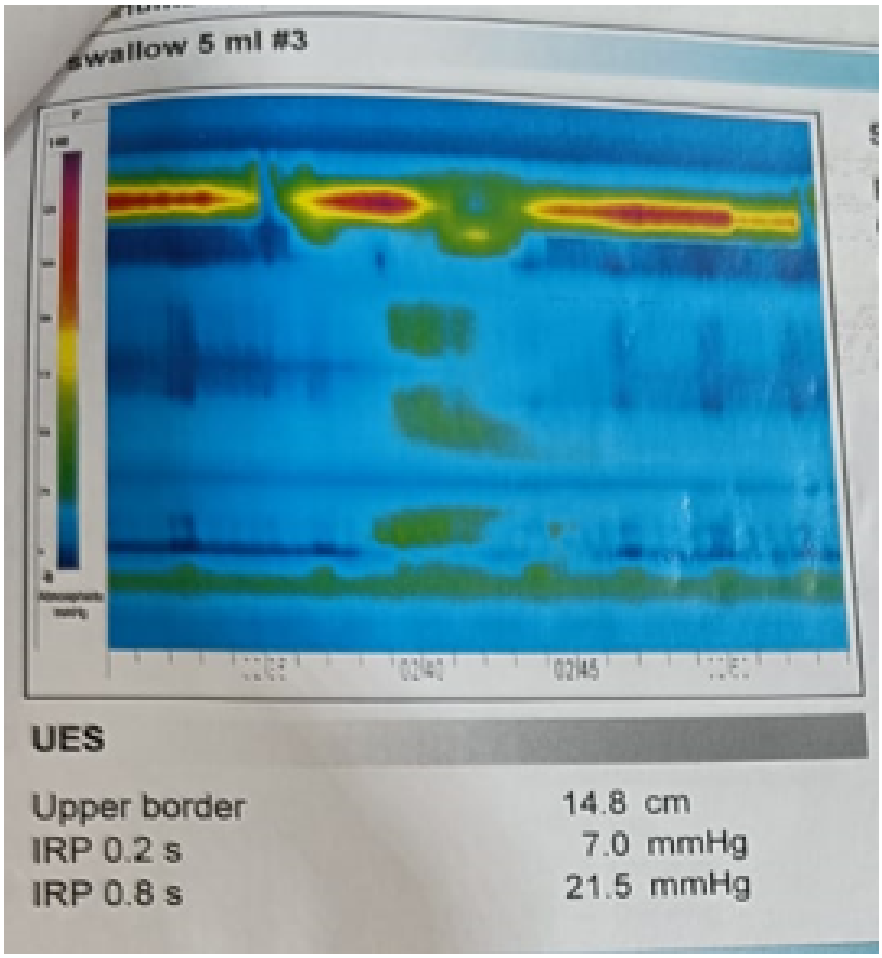
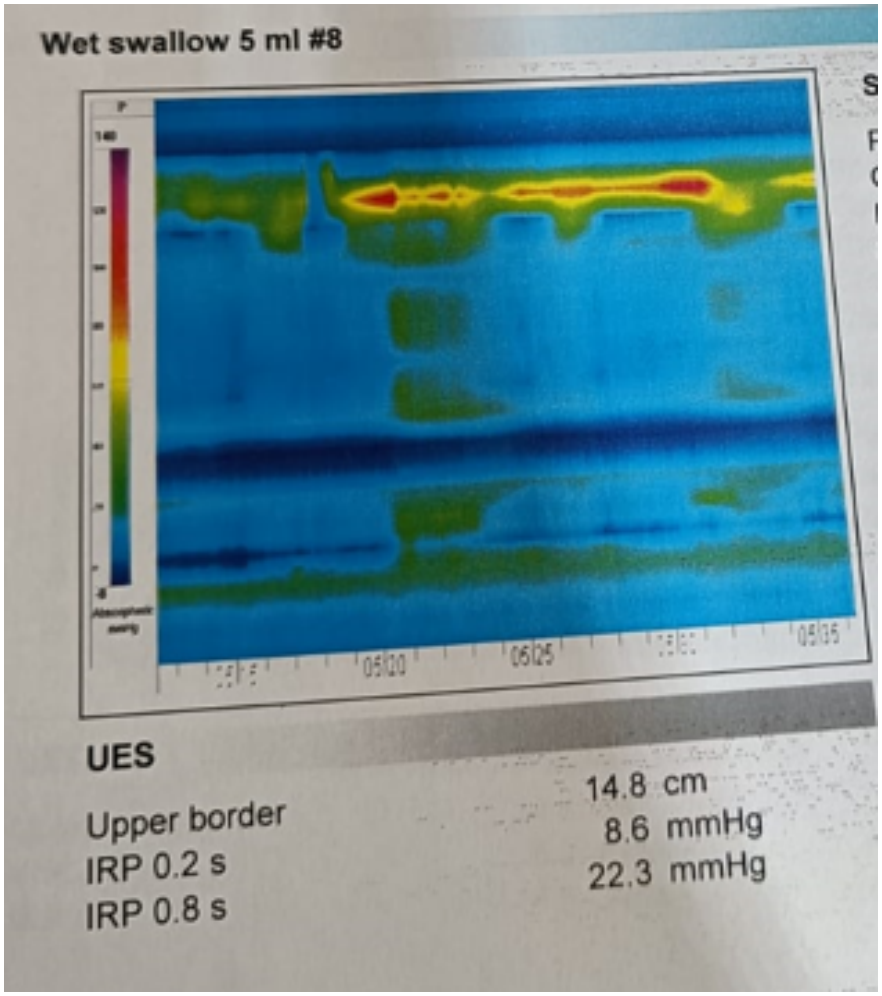


Fig -1 : Barium esophagogram showing narrowing at the distal thoracic gastroesophageal junction with dilation of the mid thoracic esophagus





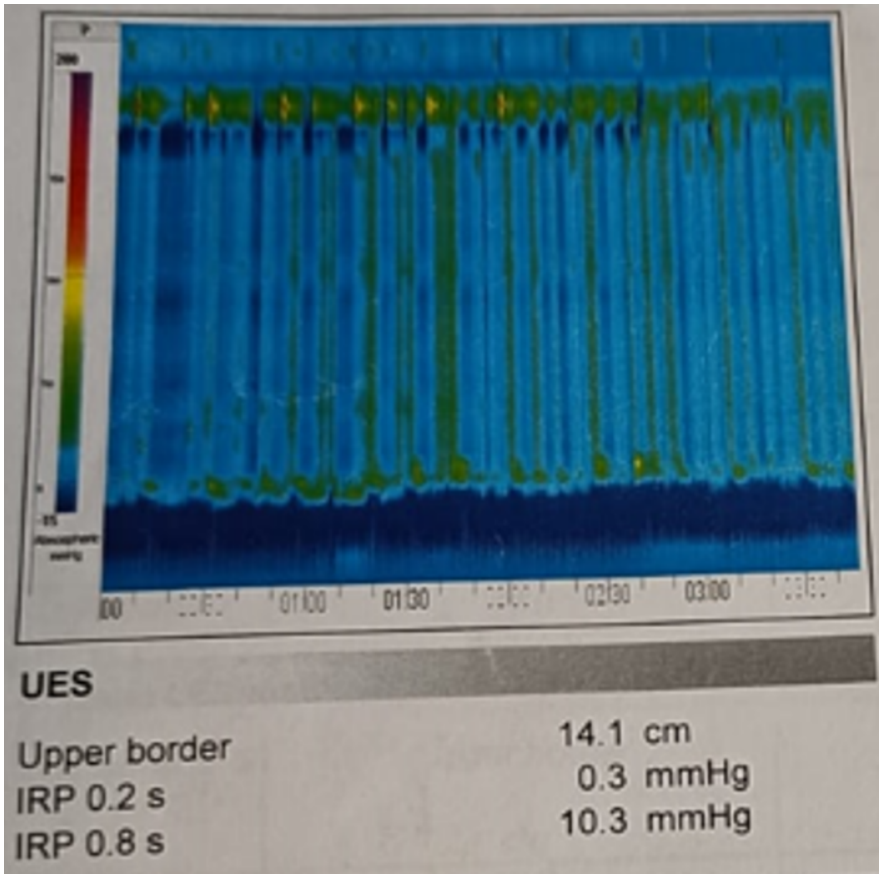
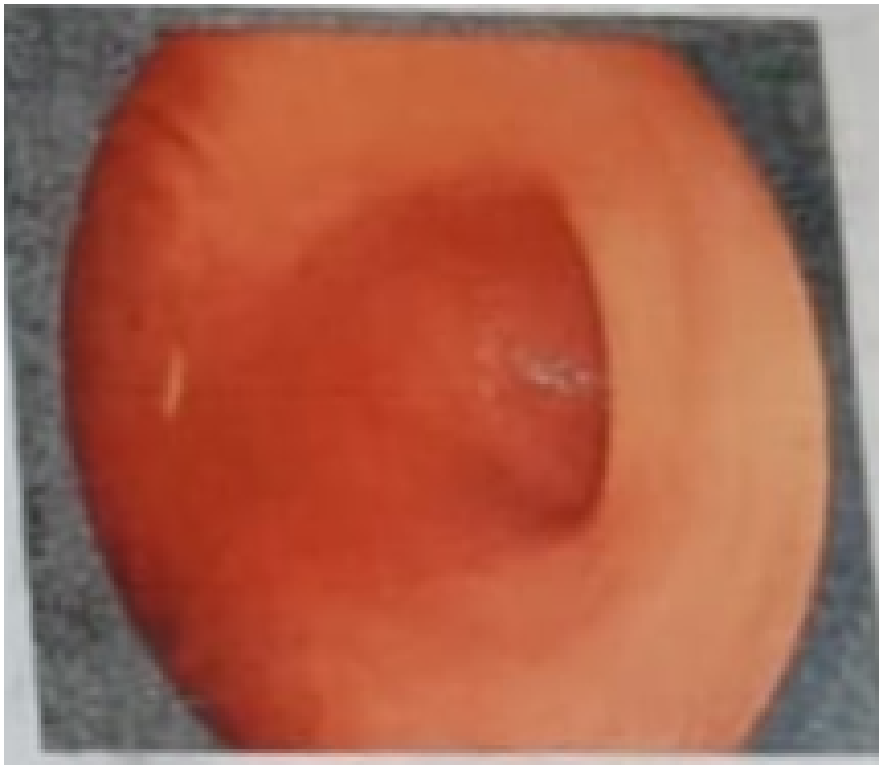


Fig-2 : Manometry showing raised Integrated Relaxation Pressure(IRP) with failed peristalsis in all swallows and pan esophageal pressurization suggestive of type II Achalasia Cardia



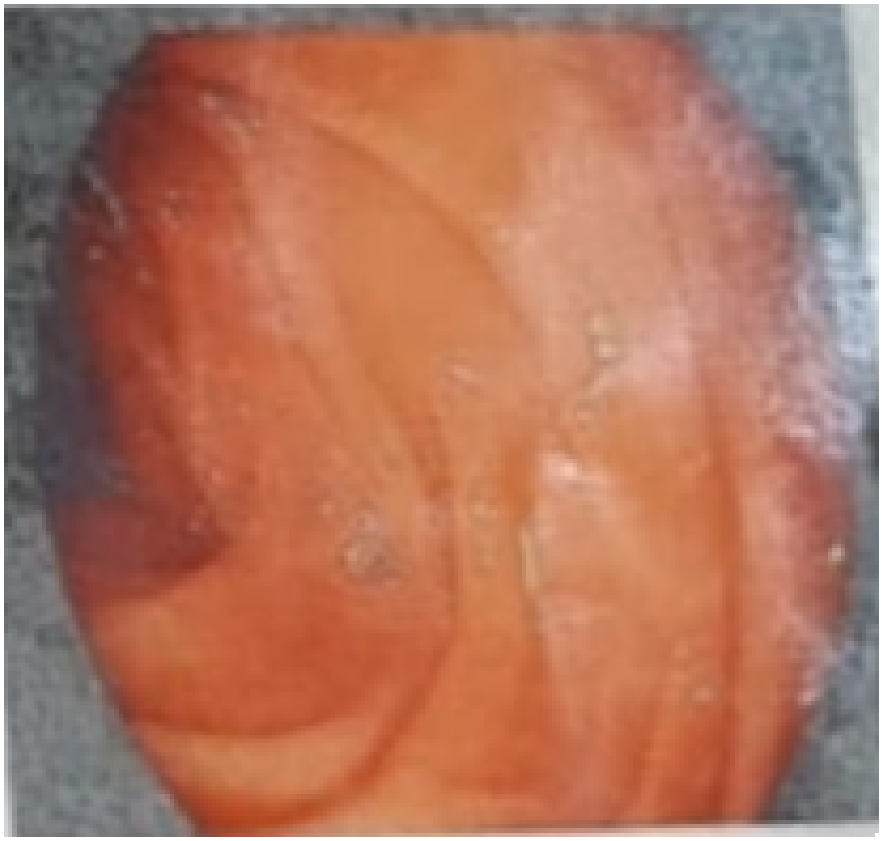
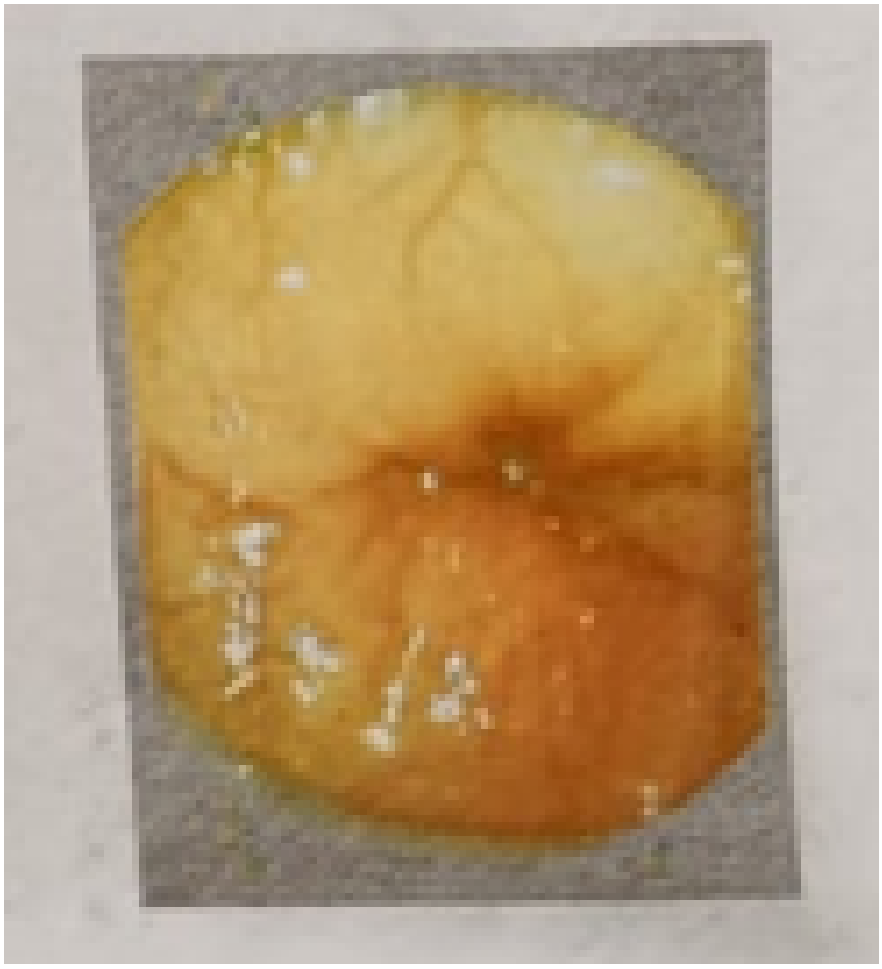
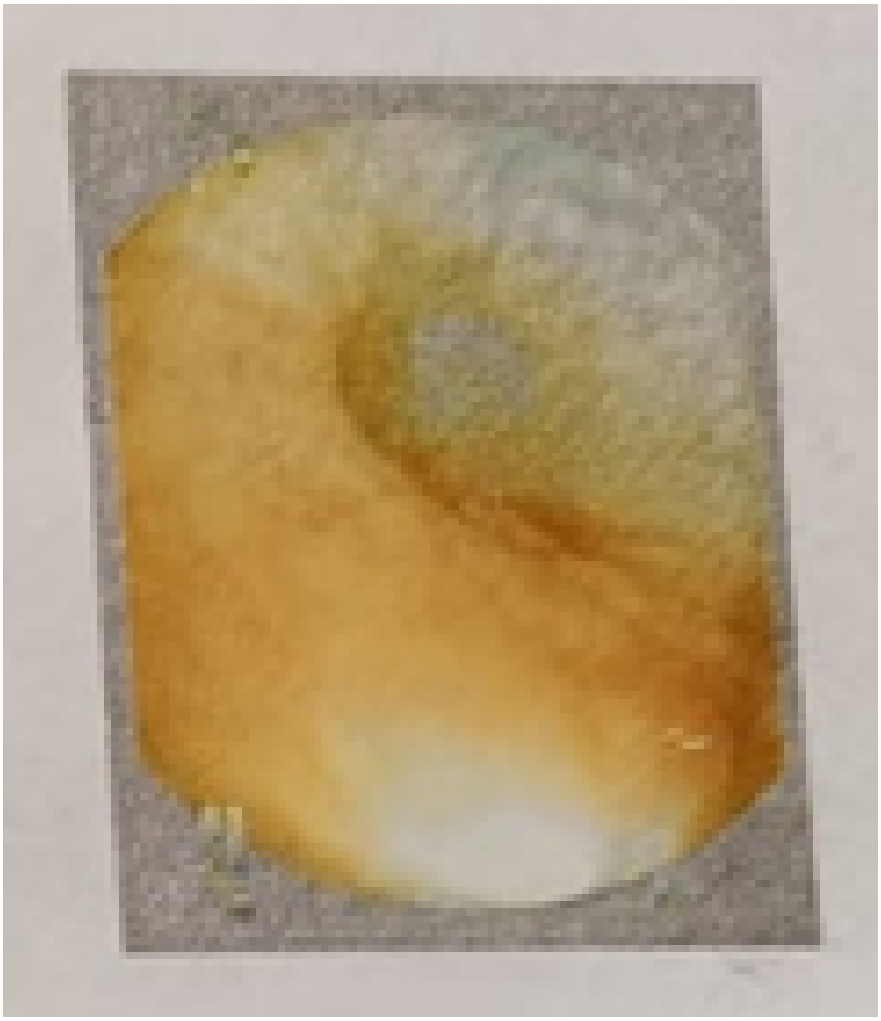


Fig-3 : Pre-dilatation upper GI endoscopy showing fundus, antrum, first(D1) and second (D2)parts of duodenum respectively





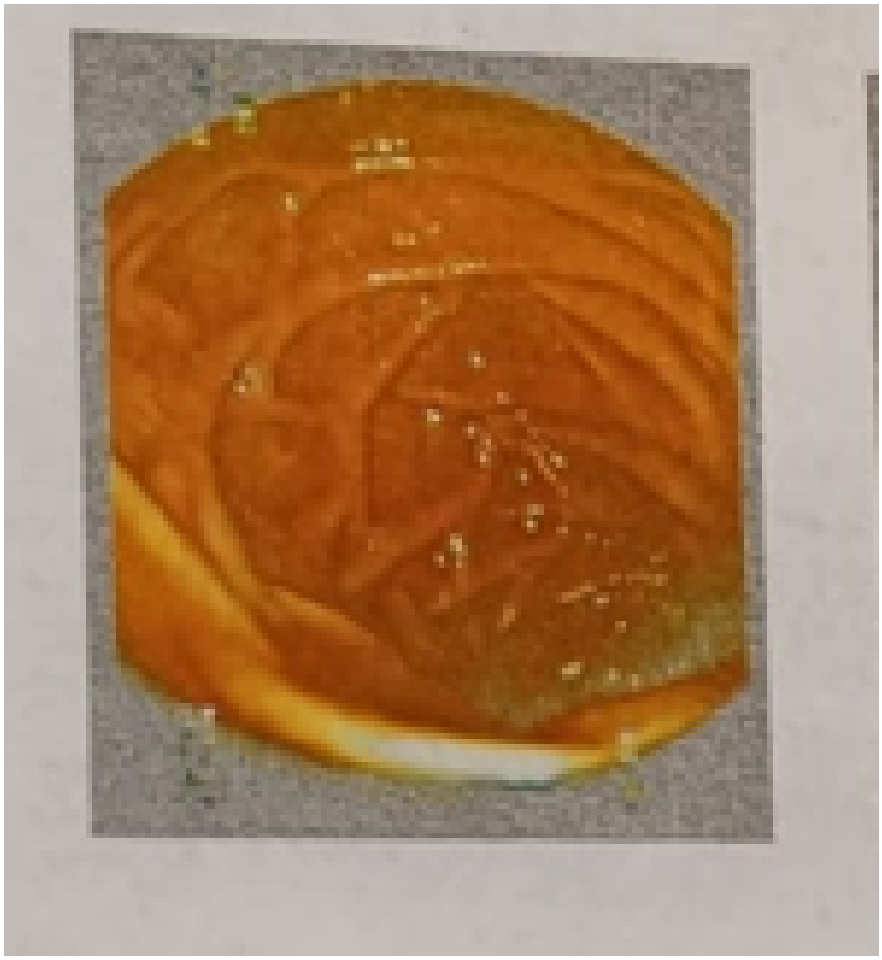


Fig-4: Post pneumatic balloon dilatation upper GI endoscopy showing fundus, antrum, first and second parts of duodenum

DISCUSSION:

The major symptom associated with achalasia is slowly progressive dysphagia for both solids and liquids and is seen in 79%-100% of affected patients. Other common symptoms are regurgitation (63%), heartburn (41%), non-cardiac chest pain (22%), epigastric pain (15%) and odynophagia (<5%). In patients with achalasia, respiratory symptoms are also common: cough (37%), aspiration (31%), hoarseness (21%), wheezing (16%), sore throat (12%), dyspnea (10%) and weight loss (10%) (Table I).[13,14]

Symptoms	**Frequency**
Dysphagia	79-100%
Regurgitation	63%
Heartburn	41%
Cough	37%
Aspiration	31%
Non cardiac chest pain	22%
Hoarseness	21%
Wheezing	16%
Epigastric Pain	15%

Sore Throat	12%
Dyspnea	10%
Weight loss	10%
Odynophagia	<5%

Table I: Symptoms associated with Achalasia Cardia

Studies have emphasized the difficulties in diagnosing achalasia, especially in young adults, due to its diverse presentation and resemblance to other esophageal conditions.[15] Early identification remains a challenge as achalasia is rare and is often diagnosed late. In addition to this, its symptoms are frequently mistaken for GERD, leading to delayed diagnosis. Misdiagnosis may result in prolonged symptom duration which necessitates additional diagnostic assessments.[16]

The patient in this case study, a 22-year-old woman, had symptoms of dysphagia and regurgitation. She was initially diagnosed with gastroesophageal reflux disease and treated with antacid gel and a proton pump inhibitor for two years , but these treatments did not alleviate her symptoms. Subsequent tests, including manometry, a barium esophagogram, and a gastroscopy, suggested a diagnosis of Type II Achalasia cardia.[12] Diagnosis of achalasia relies on clinical symptoms and imaging studies like barium swallow and esophageal manometry.[8] A barium esophagogram is the best initial test, showing classic findings such as the "bird's beak" appearance, and esophageal dilation.[9] Esophageal manometry is the most sensitive test and remains the gold standard, with high-resolution manometry being the preferred method which shows absent peristalsis.[10] Upper GI endoscopy is recommended to exclude malignancy

The objective of achalasia therapy is to alleviate symptoms by eliminating outflow resistance, which is caused by the hypertensive and non-relaxing lower esophageal sphincter (LES).[17] Both non-surgical options like pharmacotherapy and Botox injection, and surgical techniques such as pneumatic dilation, laparoscopic Heller myotomy, and per-oral endoscopic myotomy (POEM) are available treatments for achalasia.[11] Only about 10% of patients benefit from pharmacological treatment and it is typically used in elderly patients who are not suitable for pneumatic dilatation or surgery. Endoscopic treatment involves injecting botulinum toxin into the LES to block acetylcholine release and restore the balance between excitatory and inhibitory neurotransmitters. However, this treatment has limited value, and only about 30% of patients experience relief of dysphagia one year after treatment. Most patients require repeated botulinum toxin injections.[18] Pneumatic dilatation, performed by a qualified gastroenterologist, is the recommended treatment for sporadic cases where surgery is not appropriate.[19] In our case, the patient was given the option of pneumatic dilatation or Heller's myotomy. She had chosen pneumatic dilatation over Heller's myotomy. The PBD procedure commenced with the patient sedated with continuous vital signs monitoring. After balloon dilation, post-procedure examination showed improved LES function without significant complications.

The patient showed clinical improvement with relief of dysphagia, regurgitation, and heartburn symptoms after the treatment and was discharged. She was advised to adopt lifestyle changes, like eating small meals while upright to aid food passage by gravity and avoiding lying flat but instead maintaining an angle of 30 to 45 degrees to reduce the risk of aspiration. The patient was planned to follow up after 6 months with upper GI endoscopy and manometry in the outpatient clinic. Regular endoscopic surveillance is necessary in this case as there is a risk of transformation to esophageal carcinoma.[20] It's important to recognize that current treatments for achalasia are aimed at easing symptoms like regurgitation, chest pain, and difficulty swallowing, as well as preventing complications such as megaesophagus, weight loss, and gastroesophageal reflux disease.[21]

CONCLUSION

This case report emphasizes the importance of considering achalasia as a differential diagnosis in young adults presenting with dysphagia, particularly when symptoms are refractory to conventional treatments for acid peptic disorder. Prompt diagnosis and appropriate intervention, such as pneumatic dilation, can lead to significant clinical improvement and better patient outcomes.

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CONFLICT OF INTEREST

The authors report no conflicts of interest.

DATA AVAILABILITY STATEMENT

No data were used.

CONSENT STATEMENT

Written informed consent was obtained from the patient to publish this report in accordance with the journal's patient consent policy.

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