




CASE REPORT

Achalasia cardia: A case report in young female

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Key Clinical Message

This case emphasizes the need for early recognition and accurate diagnosis of achalasia in young adults to avoid exacerbation of the condition and misdiagnosis as GERD. Patient outcomes and quality of life are greatly enhanced by suitable diagnostic techniques, appropriate therapy, interdisciplinary care, and comprehensive patient education along with frequent follow-ups.

Abstract

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 symptoms include progressive dysphagia for solids and liquids, along with regurgitation, heartburn, and non-cardiac chest pain. 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 6 months with upper gastrointestinal (UGI) endoscopy and manometry in the outpatient clinic for regular endoscopic surveillance as there is a risk of transformation to esophageal carcinoma. 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. UGI 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,

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and per-oral endoscopic myotomy (POEM). The treatment options depend upon the patient's condition at presentation and their individual choices. 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, case report, dysphagia, manometry, pneumatic dilatation

1 | 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 gastrointestinal (UGI) endoscopy is further recommended to exclude malignancy.¹⁰ Treatment strategies encompass both non-surgical options like pharmacotherapy (calcium channel blockers, nitrates, and botulinum toxin) and surgical techniques such as pneumatic balloon dilation (PBD), laparoscopic Heller’s myotomy (LHM), 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.

2 | CASE HISTORY AND EXAMINATION

A 22-year-old female patient first presented to the outpatient department (OPD), with a 1-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 3 months. She reported experiencing similar symptoms intermittently over the previous 2 years and her condition did not improve with antacid treatment. The patient had no reported allergies and is not currently taking any medications. Importantly, there was 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.

3 | METHODS (DIFFERENTIAL DIAGNOSIS, INVESTIGATIONS AND TREATMENT)

Acid peptic disease with gastroesophageal reflux, diffuse esophageal spasm, esophageal stricture, and achalasia cardia are important differential diagnoses for the patient’s symptoms. To rule out acid peptic disease, an upper gastrointestinal (UGI) endoscopy was first performed, which revealed dilation in the lower portion of the esophagus with normal mucosa and absent varices. No abnormalities were detected in the gastric fundus, gastric antrum, and first and second parts of the duodenum (Figure 1A–D). To further investigate

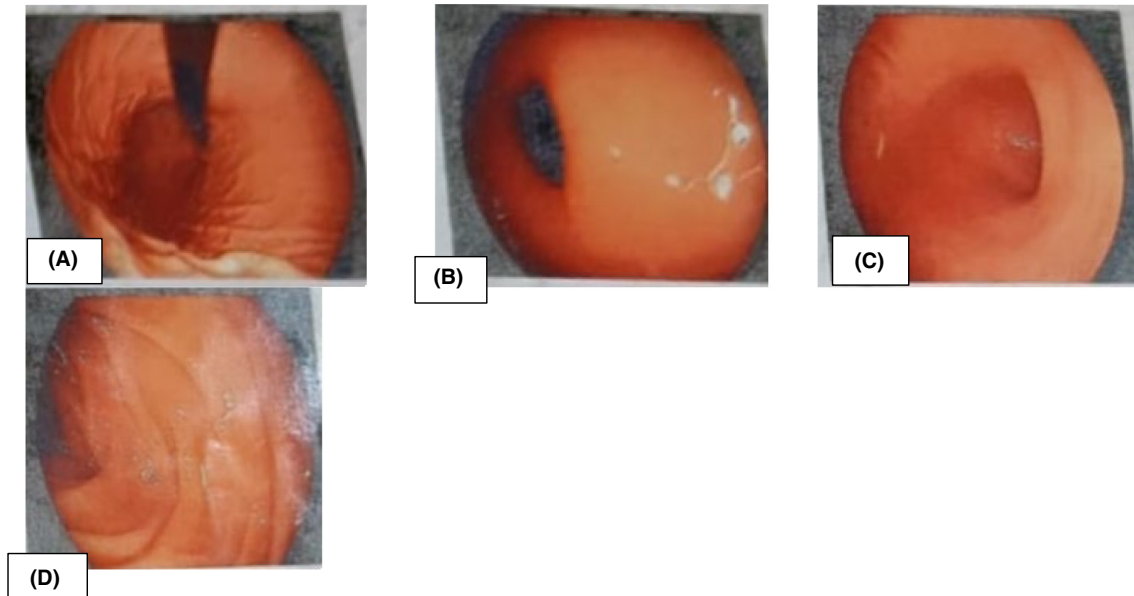


FIGURE 1 Pre-dilatation upper gastro-intestinal endoscopy showing normal fundus (A), antrum (B), first (C) and second (D) parts of duodenum respectively.

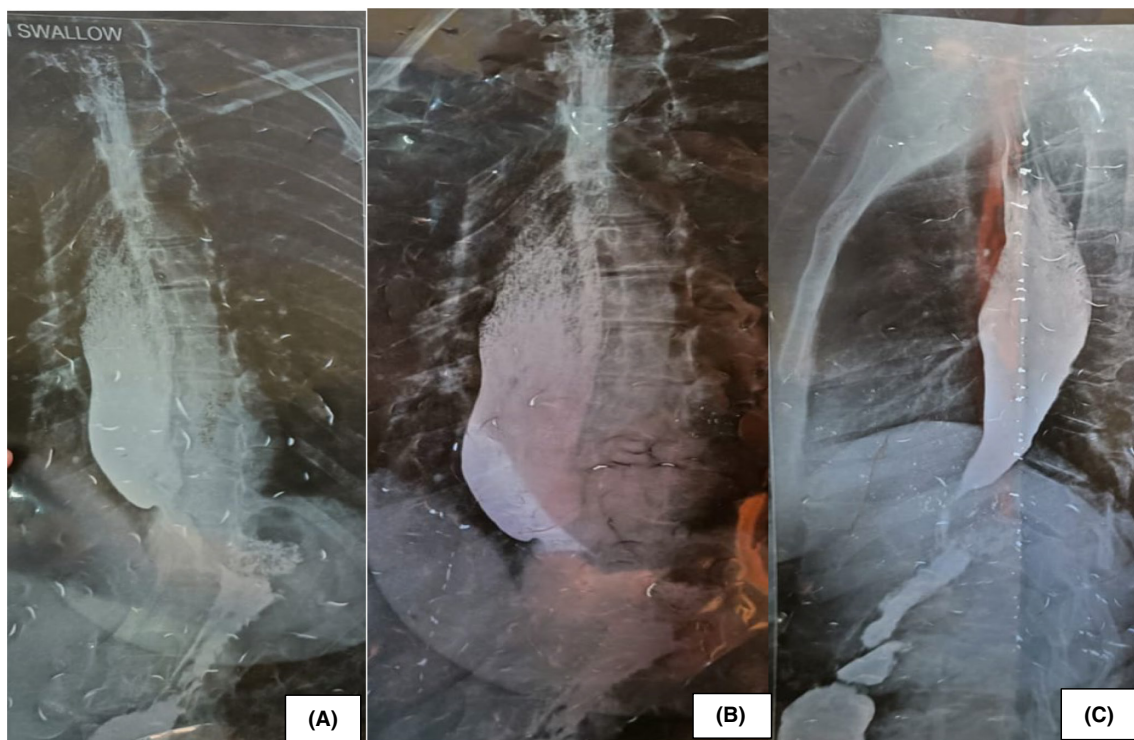


FIGURE 2 (A, B) Barium esophagogram showing dilation of the mid thoracic esophagus. (C) Barium esophagogram showing narrowing at the distal thoracic gastroesophageal junction.

potential motility disorders, a barium esophagogram, and manometry were scheduled. A barium esophagogram performed the next day 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 (Figure 2A–C). Manometry confirmed ineffective esophageal motility showing raised

Integrated Relaxation Pressure (IRP) with incomplete relaxation with failed peristalsis in all swallows and pan esophageal pressurization suggestive of type II Achalasia Cardia (Figure 3A–C). The patient was thoroughly explained about the diagnosis, available treatment options, and potential complications if left untreated. She was prescribed pantoprazole 40 mg per oral once daily, ondansetron 4 mg

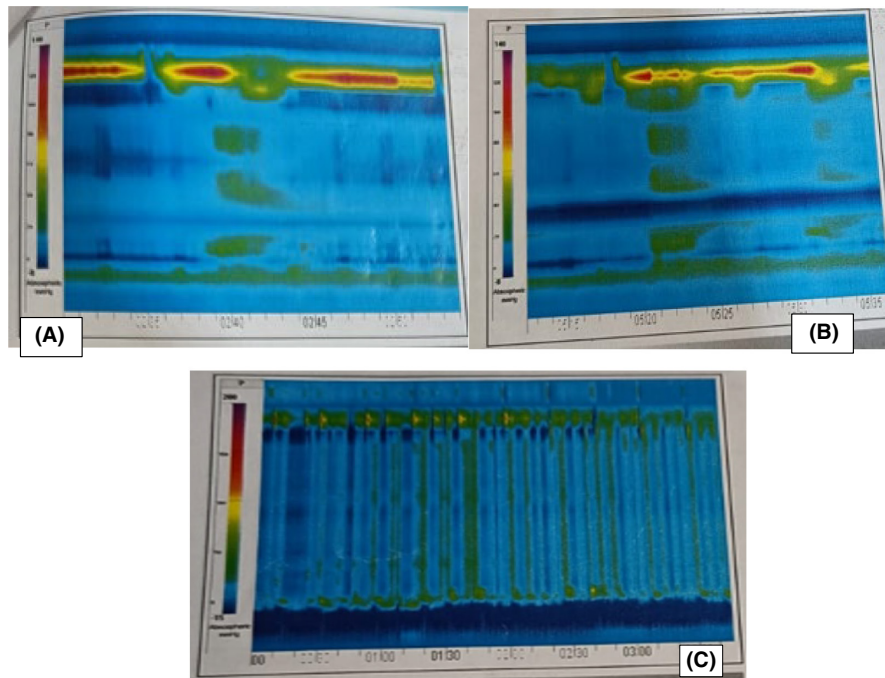


FIGURE 3 (A, B) Manometry showing failed peristaltic integrity and absent contraction pattern. (C) Manometry showing raised Integrated Relaxation Pressure (IRP) with incomplete relaxation with failed peristalsis in all swallows and pan esophageal pressurization suggestive of type II Achalasia Cardia.

as needed, and sucralfate 10 mL per oral thrice a day for 2 months. Since advanced surgical techniques such as pneumatic balloon dilatation (PBD) were not accessible at that facility, the patient was referred to a tertiary center for additional treatment.

About 6 months after diagnosis, the patient and her guardian sought care at a tertiary center. During these 6-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 PBD or LHM, accompanied by comprehensive discussions outlining the advantages and disadvantages of each procedure. She opted for PBD and was admitted the next day of her visit, where she followed a clear liquid diet regimen before undergoing the procedure 2 days later.

The PBD procedure was done by sedating the patient with midazolam (15 mcg/kg, intravenous) and propofol (2 mg/kg, intravenous). Her vital signs were monitored throughout the procedure by the team of anesthesiologists and gastroenterologists performing it. UGI endoscopy revealed a dilated esophagus with a relatively tight LES but no obvious stricture. A foreign body of meat bolus was found and removed with a dormia basket. No abnormality were detected in the second part of the Duodenum (D2) and fundus, and pseudo-achalasia was 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 pounds per square inch (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 (Figure 4A) and no obvious tear or active bleeding (Figure 4B–E). 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.

4 | OUTCOME AND FOLLOW-UP

The patient showed clinical improvement with relief of dysphagia, regurgitation, and heartburn symptoms after the treatment and was discharged. She was counseled to adopt lifestyle changes, to aid in symptom management, such as consuming small meals while remaining upright to facilitate food passage by gravity, avoiding lying flat, and maintaining an angle of 30–45° to reduce the risk of aspiration. She was advised to consume adequately chopped and minced semisolid foods, accompanied by plenty of fluid intake. Additionally, she was counseled to avoid oily and spicy foods, as well as tea and coffee, as these can exacerbate symptoms. The patient was also informed that achalasia is a chronic condition and that treatment outcomes are focused on symptom relief rather than a complete cure. A follow-up appointment was scheduled 6 months later, involving UGI endoscopy and manometry

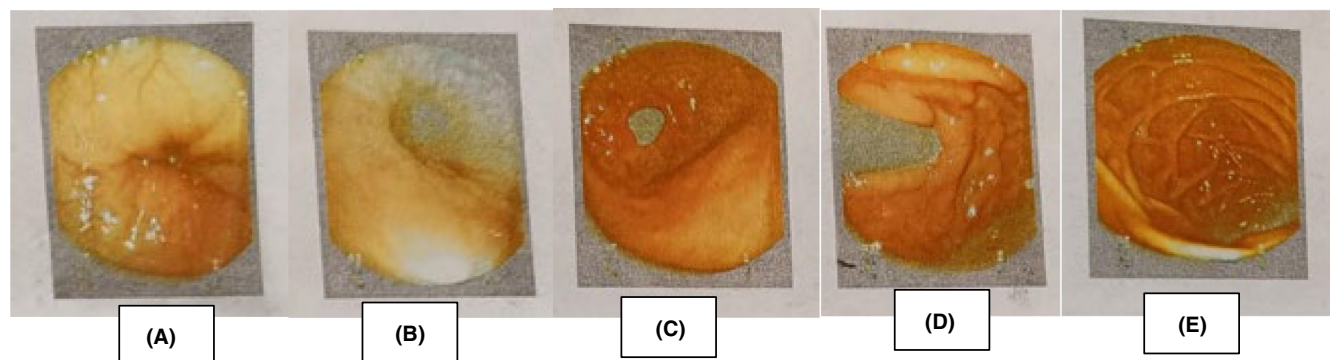


FIGURE 4 (A) Upper gastrointestinal endoscopy showing slightly dilated esophagus with minimal resistance on passing the endoscope. (B–E) Post-dilatation upper gastro-intestinal endoscopy showing fundus (B), antrum (C), first (D) and second (E) parts of duodenum respectively with no obvious tear or active bleeding.

in the outpatient clinic. She was also advised to undergo regular endoscopic surveillance every 3 years due to the risk of progression to esophageal carcinoma.

In young adults with achalasia, effective treatments like PBD, LHM, and POEM improve daily living by improving esophageal function and restoring normal eating and drinking abilities. It significantly enhances psychological well-being, along with reducing anxiety and depression associated with chronic dysphagia, despite occasional challenges such as disease progression or the need for re-treatment.

5 | 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 1).^{6,7}

Studies have emphasized the difficulties in diagnosing achalasia, especially in young adults, due to its diverse presentation and resemblance to other esophageal conditions.¹² 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.¹³ Although achalasia cardia is rare in young adults, there are few studies that reported the diagnosis in young adults (Table 2).

The patient in this case study, a 22-year-old woman, had symptoms of dysphagia and regurgitation. She was

TABLE 1 Symptoms associated with Achalasia Cardia.

Symptoms	Frequency
Dysphagia	79%–100%
Regurgitation	63%
Heartburn	41%
Cough	37%
Aspiration	31%
Noncardiac chest pain	22%
Hoarseness	21%
Wheezing	16%
Epigastric pain	15%
Sore throat	12%
Dyspnea	10%
Weight loss	10%
Odynophagia	<5%

initially diagnosed with gastroesophageal reflux disease and treated with antacid gel and proton pump inhibitors for 2 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 according to the Chicago Classification (Table 3).^{9,24} Diagnosis of achalasia relies on clinical symptoms and imaging studies like barium esophagogram 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 UGI endoscopy is recommended to exclude malignancy.

The objective of achalasia therapy is to alleviate symptoms by eliminating outflow resistance, which is caused

TABLE 2 Cases of achalasia in young adults along with intervention done and outcome.

Study	Age at diagnosis (years)	Symptoms	Intervention	Outcome
Achalasia with megaesophagus and tracheal compression in a young patient: A case report ²¹	23	Dysphagia, significant weight loss and respiratory distress	Laparoscopic Heller's myotomy with anterior semi-fundoplication	Improved
Achalasia: A case report on its effect during surgical decision making for laparoscopic sleeve gastrectomy in the young morbidly obese patient ²²	23	Dysphagia, regurgitation	Per-oral endoscopic myotomy	Improved
Esophagus achalasia: differential diagnosis of asthma ²³	19	Dyspnea, cough, vomiting	Heller's Myotomy	Improved

Type	Lower esophageal sphincter	Esophageal body
I	Incomplete relaxation	Aperistalsis and absence of esophageal pressurization
II	Incomplete relaxation	Aperistalsis and panesophageal pressurization in at least 20% of swallows
III	Incomplete relaxation	Premature (spastic) contractions with distal contractility integral (DCI) >450 mmHg·s·cm with ≥20% of swallows

TABLE 3 Manometric Chicago classification for Achalasia.

by the hypertensive and non-relaxing LES.¹⁴ Both non-surgical options like pharmacotherapy and Botox injection and surgical techniques such as PBD, LHM, and POEM are available treatments for achalasia.¹¹ Oral pharmacologic treatments are the least effective for achalasia, offering only temporary relief and often causing side effects like headaches, hypotension, and pedal edema. They are usually reserved for elderly patients who cannot undergo more definitive treatments like pneumatic dilation, or surgical myotomy as well as for those who have not responded to botulinum toxin injections.²⁵ 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, as only about 30% of patients experience relief of dysphagia 1 year after treatment, and most patients require repeated botulinum toxin injections.¹⁵ The success of each treatment option depends on various factors, such as the type of procedure (surgical methods are generally more effective than medical interventions), the severity of the disease at diagnosis (better outcomes in early-stage achalasia, worse in advanced cases), and patient-specific factors such as age and concurrent health conditions.^{16,19}

Both PBD and LHM exhibit comparable success rates and post-treatment quality of life.^{20,25} However, PBD is the most common first-line treatment for achalasia, targeting the circular muscle fibers of the LES to relieve obstruction, because of its cost-effectiveness, and less invasive

nature.^{16,19} Post-intervention, achalasia patients usually experience significant relief from dysphagia, regurgitation, and chest pain, leading to improved quality of life. They need dietary modifications and regular follow-ups to monitor for recurrence.^{17,26} The recurrence rates for achalasia treatment are 20%–30% for PBD, 10%–20% for LHM, and favorable but pending long-term data for POEM as it is a recent advancement in treatment.²⁸ The long-term goals of current treatments are to mitigate symptoms such as regurgitation, chest pain, and dysphagia, as well as prevent complications like megaesophagus, weight loss, and gastroesophageal reflux disease.^{18,27} Furthermore, regular endoscopic surveillance is needed due to the potential risk of progression to esophageal carcinoma.¹⁷

In this particular case, pharmacotherapy was deemed unsuitable due to the patient's young age, the transient nature of symptom relief, and the associated side effects. Botulinum toxin injection was also considered of limited value, given that only a small percentage of patients experience sustained relief after 1 year, and repeated treatments are often required. Thus, the patient was given the option of PBD or LHM. However, in our case, PBD was chosen due to the patient's preference, its cost-effectiveness, its less invasive nature, and the local expertise of the treating physicians. The PBD procedure commenced with the patient being sedated with continuous vital signs monitoring. After balloon dilation, the post-procedure examination showed improved LES function without significant complications.

6 | 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 APD. Despite advancements in treatment, our understanding of the pathophysiology of achalasia remains incomplete. Although POEM represents a promising recent treatment, its application is constrained by the availability of highly skilled practitioners and should be reserved for high-volume centers with experienced operators. Further research and ongoing monitoring are necessary to deepen our understanding of the pathophysiology of disease and to evaluate long-term outcomes.

AUTHOR CONTRIBUTIONS

Deepak Subedi: Conceptualization; data curation; investigation; methodology; resources; supervision; writing – original draft; writing – review and editing. **Binod Raj Parajuli:** Data curation; investigation; methodology; project administration; resources; writing – original draft. **Neha Bista:** Conceptualization; data curation; investigation; resources; supervision; writing – original draft; writing – review and editing. **Somee Rauniyar:** Investigation; resources; supervision; writing – review and editing. **Kiran Dhonju:** Supervision; writing – original draft; writing – review and editing. **Santosh Bhusal:** Investigation; resources; writing – review and editing. **Egesh Aryal:** Supervision; writing – review and editing. **Divas Adhikari:** Supervision; writing – original draft; writing – review and editing. **Saurav Aryal:** Supervision; writing – review and editing. **Ayush Karna:** Writing – review and editing.

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CONFLICT OF INTEREST STATEMENT

The authors report no conflicts of interest.

DATA AVAILABILITY STATEMENT

Data sharing not applicable—no new data generated, or the article describes entirely theoretical research.

CONSENT

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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