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CASE REPORT

Life-threatening end-stage achalasia – a case report of neglected dysphagia

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Achalasia is a rare oesophageal dysmotility disorder characterised by the absence of peristalsis and impaired lower oesophageal sphincter (LOS) relaxation. Delayed diagnosis is common, often due to the misinterpretation of symptoms for more common conditions like gastro-oesophageal reflux, leading to significant morbidity and reduced quality of life. This case emphasises the importance of diagnosing achalasia timeously and highlights diagnostic modalities and therapeutic options currently used.

A 37-year-old patient with a 12-year history of progressive dysphagia, weight loss, and severe dental decay presented after a syncopal episode. The workup revealed severe dehydration, refractory hyperkalaemia, and renal failure requiring haemodialysis. Further investigations demonstrated advanced achalasia with a dilated sigmoid oesophagus. High-resolution manometry (HRM) confirmed type I classic achalasia. The patient underwent a laparoscopic Heller myotomy (LHM) after nutritional stabilisation.

This case underscores the difficulty of diagnosing achalasia, especially in its early stages, where symptoms may be mistaken for reflux disease. Misdiagnosis delays appropriate treatment, leading to significant morbidity, including malnutrition. Dental erosions may also occur due to the frequent regurgitation of fermented food. A high index of suspicion coupled with proper diagnostic investigations, such as HRM and imaging, is essential for timely diagnosis and management. Surgical options like LHM remain the gold standard treatment for type I achalasia.

Achalasia, though rare, should be considered in patients with unexplained dysphagia and associated symptoms. A timely diagnosis can prevent complications such as malnutrition and oesophageal damage. Physicians must recognise atypical presentations and consider HRM in cases with refractory symptoms to ensure early intervention and optimal outcomes.

Keywords: end-stage achalasia, dysphagia, high-resolution manometry

Case report

A 37-year-old man was brought to the Emergency Department of Groote Schuur Hospital, Cape Town, South Africa, by ambulance after a syncopal collapse. On arrival, he was conscious with a Glasgow Coma Scale score of 15/15, although markedly lethargic. Blood pressure was 169/140 mmHg, with a heart rate of 78 beats per minute, a temperature of 35 °C, a respiratory rate of 16 breaths per minute, and a finger-prick glucose of 9.8 mmol/L. He was given intravenous (IV) fluid and, on further history, complained of extreme thirst, lethargy, progressive dysphagia, and weight loss of more than 40 kg. He had no known chronic medical comorbidities, was not taking any chronic medications, was not known to have any allergies, and had no prior surgery.

On examination, he was noted to be dehydrated and mildly wasted, but with normal respiratory, cardiovascular, and abdominal examinations. He was noted to have grossly carious dentition, with decay of all his teeth (Figure 1). Arterial blood gas (on room air) demonstrated pH: 7.34, pO_2: 17.2 kPa, pCO_2: 3.15 kPa, bicarbonate: 17.1 mEq/L, base excess: -12.9, and potassium:

Figure 1: Severe dental decay noted to involve both upper and lower teeth, but more severe on the posterior aspect of the teeth.
a.) Anterior teeth (clenched together)
b.) Upper teeth showing predominant posterior tooth decay.
c.) Lower teeth showing predominant posterior tooth decay

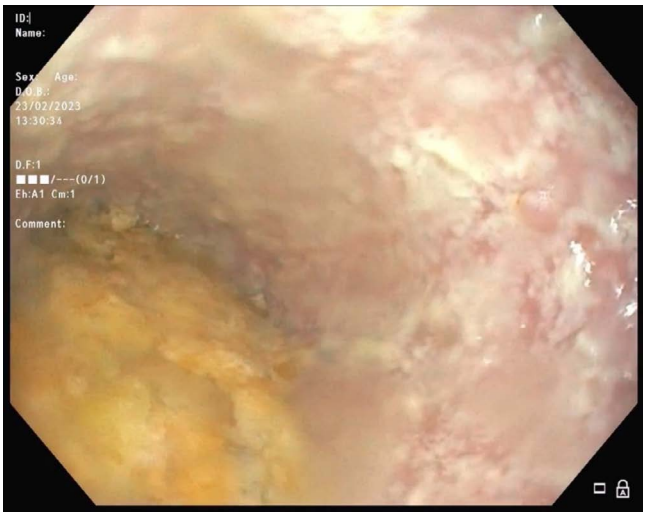

Figure 2: Endoscopic view of oesophagus in end-stage achalasia with significant dilatation, retained food and fluid, and severe stasis oesophagitis

6.6 mmol/L. Formal serum blood investigations showed sodium: 134 mmol/L, potassium: 6.5 mmol/L, urea: 146.6 mmol/L, creatinine: 870 µmol/L, phosphate: 4.03 mmol/L, magnesium: 1.54 mmol/L, calcium: 2.14 mmol/L, white cell count: 37.5×10^9/L, haemoglobin: 21.3 g/dl, platelets: 428×10^9/L, and albumin: 48 g/L.

Immediate management and further investigations

The emergency unit instituted hyperkalaemia management, but after three cycles of 10 ml of 10% calcium gluconate and 10 units of rapid insulin with 50% dextrose, the serum potassium remained refractory at 6.4 mmol/L. Nephrology was consulted, and due to the refractory hyperkalaemia, emergency haemodialysis was initiated. He was admitted to the medical high-care unit for continuous monitoring and required two further sessions of intermittent haemodialysis before the hyperkalaemia improved. At this point, he was receiving intensive IV fluid resuscitation. A continued downward trend in his renal function and increased urine output were noted.

On day two of admission, coffee-ground vomitus was witnessed in the high care unit, and he was referred for upper gastrointestinal endoscopy. A dilated oesophagus with retained food and fluid with severe stasis oesophagitis was observed (Figure 2). The oesophagogastric junction (OGJ) was difficult to negotiate, but the endoscope could eventually be passed into the stomach, where no cardial pathology was noted on retroflexion, and no other pathology was noted distally. The findings were highly suspicious of advanced achalasia. After passing a standard hydrophilic soft-tipped 0.035" guidewire into the stomach under endoscopic vision, a large 16 Fr nasogastric (NG) tube was passed over the wire into the stomach for feeding. The patient was transferred into the care of the upper gastrointestinal surgical team.

On further, more extensive history-taking, the patient reported a 12-year history of difficulty swallowing solids. He was seen by two different general practitioners and was initially given proton pump inhibitor therapy for suspected acid reflux. Over the next four years, he found no improvement and noted that he was losing weight. After changing his diet, he was also noted to be increasingly lethargic, which resulted in him consuming greater amounts of caffeinated energy drinks. He presented to a regional hospital on four separate occasions and was eventually investigated with plain X-rays, but no further investigations or management strategies were performed.

Due to increasing concerns about missing workdays and being warned that he may lose his employment, he did not seek further

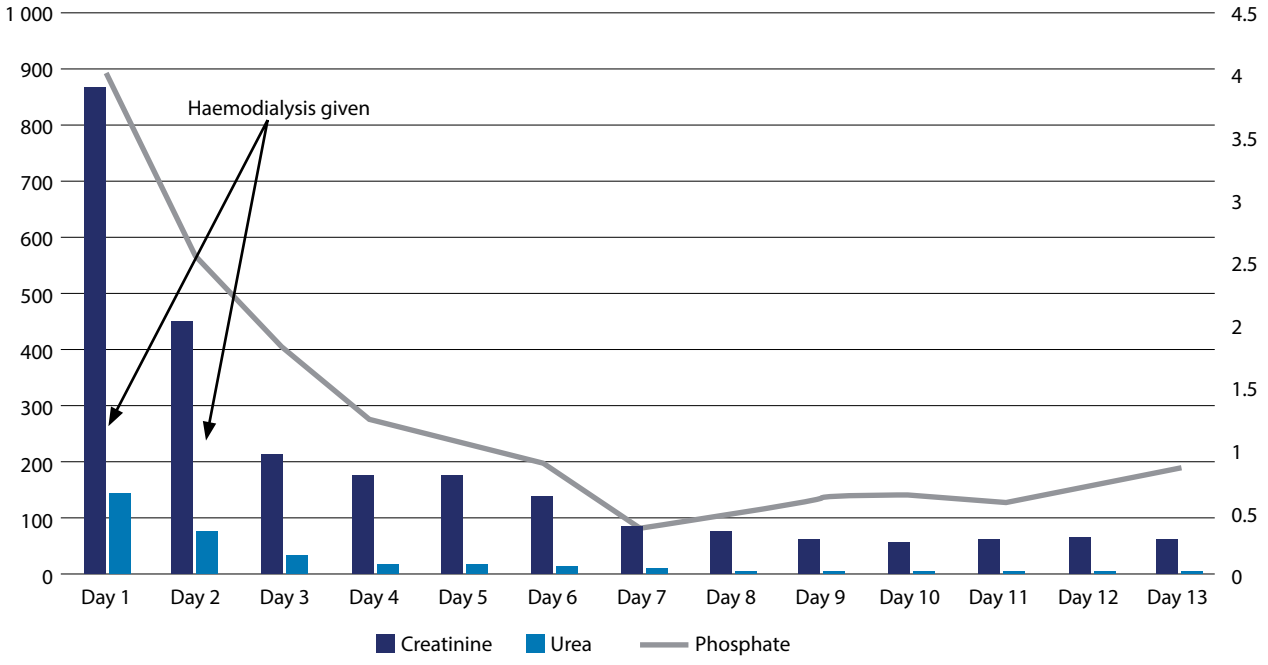

Figure 3: Trend of rapid drop in serum creatinine (µmol/L) and urea (mmol/L) over the first few days after admission until normalised by day 8, serum phosphate (mmol/L) showed a precipitous drop caused by refeeding, and hypophosphataemia dropped to a minimum of 0.35 mmol/L by day 7 despite aggressive IV phosphate replacement

healthcare for the next few years. He continued to deteriorate further with worsening dental hygiene and weight loss. When he started regurgitating even liquids, he again presented to his local community health centre, where acid reflux was again diagnosed. A week later, he collapsed while trying to buy an energy drink at his local petrol station and was brought to Groote Schuur Hospital. His Eckardt score on admission to Groote Schuur Hospital was 10/12.

Further management and special investigations

Given the presence of malnutrition and the associated high risk of refeeding syndrome, nutritional prehabilitation was initiated with a graded caloric intake of 10 kcal/kg/day via NG enteral feeding. If electrolyte levels remained stable, enteral feeding was gradually advanced by 5 kcal/kg/day. Despite this cautious approach, a rapid drop in serum phosphate was noted by the second day of enteral feeding and aggressive IV phosphate replacement was commenced. His hypophosphataemia persisted and, at its lowest level, was noted to be 0.35 mmol/L despite ongoing phosphate replacement. Once the phosphate levels normalised, the feeds were slowly increased.

Figure 3 shows the trend in serum creatinine and urea, and the subsequent rapid drop in serum phosphate over the first two weeks of admission. A total of three weeks with ongoing daily IV phosphate replacement and daily serum phosphate measurements were needed before goal feeds could be achieved. This required administering a total of > 600 mmol (> 30 amps) of IV potassium phosphate to allow for goal feeding.

During this time, a barium swallow and a computed tomography (CT) scan were performed. These showed a significantly dilated sigmoid mega-oesophagus with smooth distal tapering and a significant lack of oesophageal drainage and contrast drainage in keeping with end-stage achalasia (Figure 4). High-resolution manometry (HRM) was performed, which showed 100% absent peristalsis and increased median integrated relaxation pressure (IRP), indicating a lack of lower oesophageal sphincter (LOS) relaxation, confirming a type I classic achalasia (Figure 5).

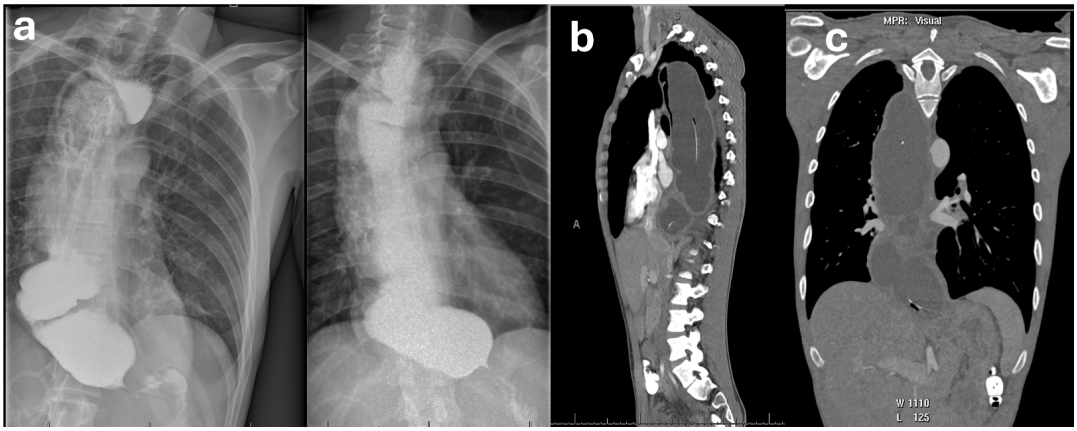

Figure 4: Barium contrast swallow and CT images
a.) Contrast swallow with two views showing significantly dilated sigmoid oesophagus, with lack of contrast drainage and smooth distal tapering at the OGJ in keeping with a "bird's beak" appearance
b.) Sagittal CT image
c.) Coronal CT image

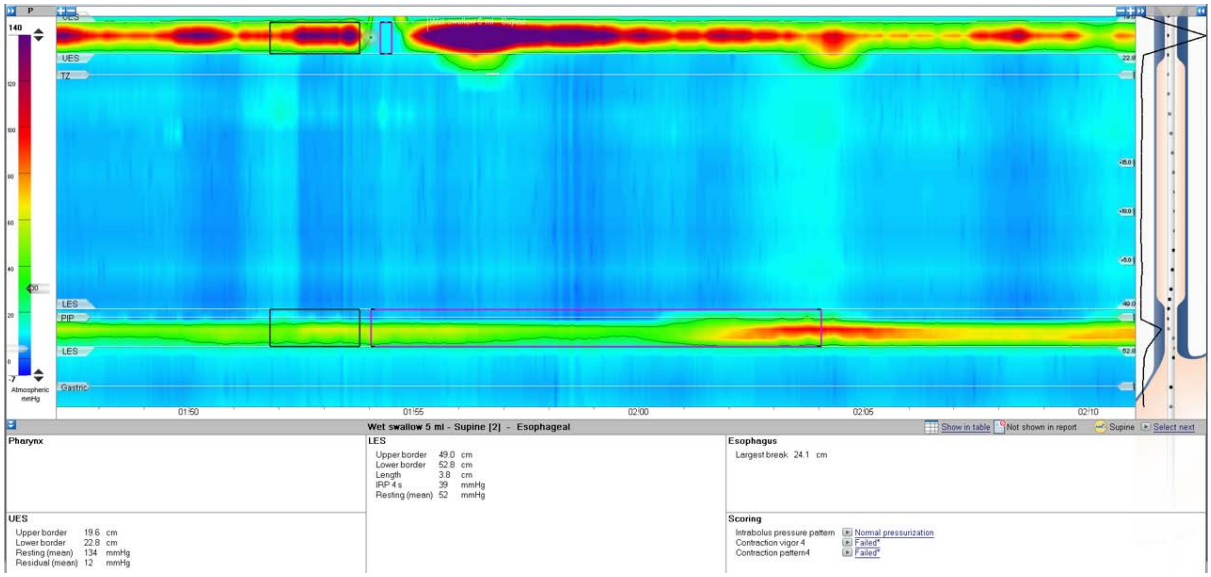

Figure 5: HRM showing single swallow with complete lack of oesophageal body peristalsis and lack of LOS relaxation; all 10 of the swallows performed looked similar, confirming type I classic achalasia

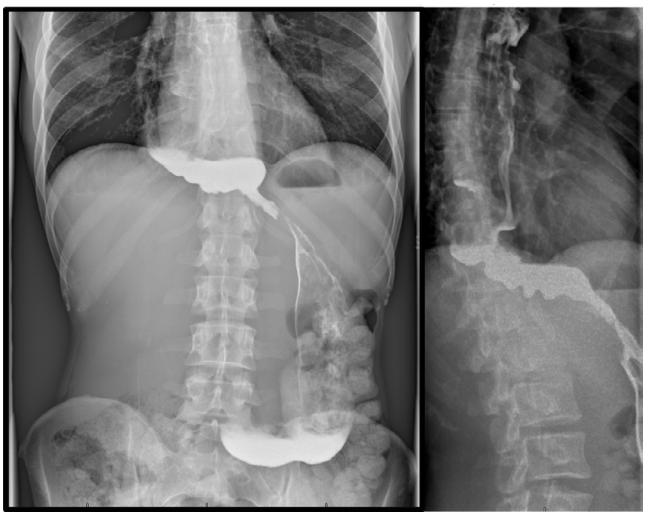

Figure 6: Postoperative contrast swallow images showing improved oesophageal drainage with reduced oesophageal diameter

Surgical management and outcome

After achieving nutritional optimisation four weeks after admission, the patient underwent a successful laparoscopic Heller myotomy (LHM). A contrast swallow examination on the first postoperative day showed significantly improved oesophageal drainage and excluded a leak from the myotomy (Figure 6). The NG tube was subsequently removed two days after the surgery, and the patient could tolerate a soft diet orally without any symptoms for the first time in over eight years. He was successfully discharged on the fourth postoperative day and will require further follow-up in the upper gastrointestinal surgery unit outpatient clinic.

Discussion

This case highlights the challenges and complexity of diagnosing achalasia timeously and the morbidity associated with a delayed diagnosis. While 12 years to diagnosis is on the extreme end of the spectrum, a delay in diagnosing achalasia remains common, with several studies demonstrating a mean time to diagnosis of approximately five years.[1,2] Several factors may account for this, including a lack of achalasia awareness in general, incorrectly identifying the symptoms of dysphagia and intra-oesophageal reflux as heartburn and gastro-oesophageal reflux, atypical symptoms early in the disease course, physician recognition and awareness of atypical presentations, patient adjustment to symptoms, and non-specific findings or findings attributable to an alternative cause on initial investigations, such as oesophagitis. Although achalasia remains a relatively rare disease, global trends suggest an increase in the overall incidence, and clinicians must be aware of both the typical and atypical presentations of this condition.[3]

Achalasia is a motility disorder of the oesophagus, resulting in aperistalsis and impaired relaxation of the LOS.[4,5] The underlying pathophysiological mechanism is the loss of myenteric neurons due to chronic ganglionitis because of a dysregulated immune response to an unknown antigen. The aetiology of this myenteric neuron loss is not fully elucidated, but various factors are thought to play a role. The hypothesis of an autoimmune condition is supported by an increased incidence of various autoimmune conditions, including type I diabetes mellitus, hypothyroidism, Sjögren syndrome, and systemic lupus erythematosus.[6] Booy et al.[7] showed that overall, patients with achalasia were 3.6 times more likely to have another autoimmune condition. The presence of antimyenteric antibodies isolated in serum supports this theory, although whether this is a reactive or inciting process has not been confirmed.[8] The role of viruses, particularly herpes simplex virus-1 (HSV-1), measles, and human papillomavirus, have also been implicated in the development of achalasia. In particular, HSV-1 has garnered attention as it is a neurotropic virus known to infect the oesophagus. Several studies have shown that latent HSV-1 causes a persistent immune response resulting in the loss of oesophageal neurons in a genetically susceptible host.[9,10]

Dysphagia, to both solids and liquids, is the hallmark of achalasia and occurs in almost all patients; however, it occurs as the first symptom in only a third of patients.[1] Other typical symptoms include regurgitation, weight loss, and retrosternal chest pain. These four symptoms comprise the basis of the Eckardt score, which is used to grade achalasia severity and assess treatment response. However, other atypical symptoms may precede the more typical symptoms and lead to alternative diagnoses in early disease. These include heartburn, epigastric pain, nausea, and, in some circumstances, extra-intestinal symptoms, like cough and aspiration.[1,2] Indeed, the diagnosis of "refractory heartburn" may even result in anti-reflux surgery. Recognition of the broader symptom set should prompt a thorough evaluation to exclude other causes of dysphagia and, ultimately, diagnose achalasia in a timely fashion.

Unfortunately, as with our patient, multiple visits to several primary care physicians failed to initiate the appropriate diagnostic workup. Typically, there are few to no clinical signs in patients with idiopathic achalasia; however, a notable clinical finding in this case was poor oral hygiene. Dental enamel erosions have been described in severe, prolonged achalasia, where the fermenting food in the oesophagus produces lactic acid. Frequent regurgitation results in demineralisation of the tooth structure with resultant dental erosions, and due to exposure occurring more frequently on the posterior aspect of the teeth, there is usually a predisposition to greater posterior than anterior carious dentition.[11,12]

The diagnosis of achalasia in our patient was ultimately made based on endoscopic, radiographic, and HRM findings, demonstrating some of the classic findings of severe achalasia. Endoscopy is usually the first investigation, as excluding a mechanical obstruction or pseudoachalasia is paramount before performing HRM. Endoscopic features supporting a diagnosis of achalasia include a dilated oesophageal body, puckered LOS, and retained saliva; however, these findings may only be noted in moderate to severe disease, as in our patient, and endoscopy might be normal early in the disease course. Besides the mega-

oesophagus, our patient had severe oesophagitis secondary to food stasis.

This case also illustrates the classic findings on a barium swallow with a "bird's beak" at the LOS and a dilated oesophagus body. A modified, timed barium swallow should also be requested, particularly in early disease, to measure the barium column one, two, and five minutes post-ingestion as an indicator of oesophageal emptying. Importantly, a normal barium swallow does not exclude achalasia. Thus, HRM should be performed in all patients with dysphagia where the endoscopy and barium swallow have not provided a diagnosis. A standard HRM protocol, as outlined by the Chicago Classification (version 4.0), helps delineate the type of motility disorder or OGJ outlet obstruction present.[13,14] Provocation manoeuvres and positional changes during the investigation can further clarify the underlying disorder. The HRM in our patient demonstrated the typical findings in type I achalasia: a raised median IRP and complete aperistalsis in all swallows.

Achalasia management is ultimately palliative, aimed at reducing symptoms, improving quality of life, and preventing long-term complications and further oesophageal dilatation by disrupting the OGJ. These can be broadly classified into pharmacological, endoscopic, and surgical therapeutic modalities. Medical management includes using calcium channel blockers, nitrates, and, less commonly, beta-adrenergic agonists and anticholinergics. However, unless a patient is not a candidate for a definitive surgical or endoscopic procedure, medical therapy is no longer recommended in achalasia due to its limited efficacy, short-term effects, and side effect profile. Endoscopic and surgical options are the first-line treatment.[15,16]

Endoscopic modalities involve mechanical disruption of the OGJ. The two main procedures are pneumatic dilatation (PD) and peroral oesophageal myotomy (POEM). Additionally, botulinum toxin can be injected into the LOS with reasonable symptom relief; however, this usually dissipates after several months, requiring repeated procedures. Most guidelines only recommend this option when patients are not good surgical candidates. PD is an effective treatment modality, with most patients having symptom relief after one year; however, this effect tends to wane over time with recurrence of symptoms in approximately 50% of patients after five years.[15-18]

Perforation occurs in about 1–2% of patients undergoing PD, and this option should only be performed in patients fit for surgery.[15] The advent of POEM has improved achalasia treatment outcomes. This is particularly the case in type III achalasia, where the length of myotomy can be tailored to involve the length of the distal contractile segment. In one randomised control trial of POEM versus PD, the two-year success rate was 92% versus 54% ($p = 0.001$).[19] The therapeutic benefit is sustained over time with less need for repeat interventions.[20]

LHM is the current surgical gold standard and is an appropriate first-line option for types I and II achalasia. It is also the treatment modality we offered our patient. His Eckardt score and symptoms improved significantly after surgery, and he could complete a meal for the first time in years. The short-term outcomes of LHM are excellent, but as in PD, the long-term effects dimmish over time: 89% at six months and 57% at six years.[21]

Gastro-oesophageal reflux disease is one of the major side effects of myotomy, and in most cases, a partial fundoplication is performed simultaneously.[15,16] In advanced achalasia, with a sigmoid or tortuous oesophagus, the above therapeutic modalities are not as effective and an oesophagectomy with gastric interposition might be necessary. However, there is significant morbidity, especially respiratory complications, and it is reserved for surgically fit patients where other treatments have failed.[15]

Conclusion

This case highlights the importance of establishing a timely diagnosis in patients with achalasia and the morbidity associated with neglected disease. Dysphagia is always an alarm symptom that should prompt a thorough investigation. Although it is a rare disease, clinicians should be aware of atypical symptoms. Where there is diagnostic doubt or no response to proton pump inhibitors in the setting of heartburn or reflux, HRM should be requested to exclude achalasia.

Conflict of interest

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Informed consent

The patient gave informed consent for the use of anonymised data and relevant photographs and images for this publication.

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