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Esophageal achalasia: A case report in an adolescent

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ABSTRACT

Esophageal achalasia is an uncommon disease in pediatrics. With an insidious clinical presentation, diagnosis is delayed.

Here we describe a case of esophageal achalasia in a 16-year-old girl, with the typical delay in consultation and diagnosis.

Although pneumatic balloon dilatation has been described as the best therapeutic option for type II achalasia, it was ineffective in our patient and she required Heller extramucosal myotomy with gastroesophageal fundoplication for reflux.

Keywords: *esophageal achalasia; dysphagia; esophageal motility disorders; manometry; Heller myotomy.*

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INTRODUCTION

Achalasia is an esophageal motility disorder of unknown etiology, rare in pediatrics.[1–6] The onset of signs and symptoms is usually insidious, secondary to progressive functional obstruction of the esophagus, presenting mainly with dysphagia and regurgitation.[1,2] The diagnosis is based on upper gastrointestinal series[1,7,8] and is confirmed with an esophageal manometry.[1,2,4,5,7,9,10] Pediatric patients are usually referred to multiple departments before seeing a gastroenterologist, especially to mental health services.[1] There are several therapeutic options, although none is curative.[1,2,5–10] Subsequent follow-up is important due to potential short- or long-term recurrence.[1–10]

Here we describe a case of esophageal achalasia in an adolescent, emphasizing the importance of a timely diagnosis for an effective treatment.

CASE REPORT

This was a 16-year-old female patient with no relevant personal or family history who consulted due to vomiting and loss of 18% of body weight (approximately 10 kg) over 6 months, without epigastric pain or heartburn. In addition to the initial vomiting, she developed dysphagia, first for solids and, in the following month, also for semi-solids and liquids.

She consulted with the Department of Ear, Nose, and Throat, where she had a fibrobronchoscopy with normal results, and with the Department of Gastroenterology, where she underwent an esophagogastroduodenoscopy, which found mild chronic gastritis due to *Helicobacter pylori*.

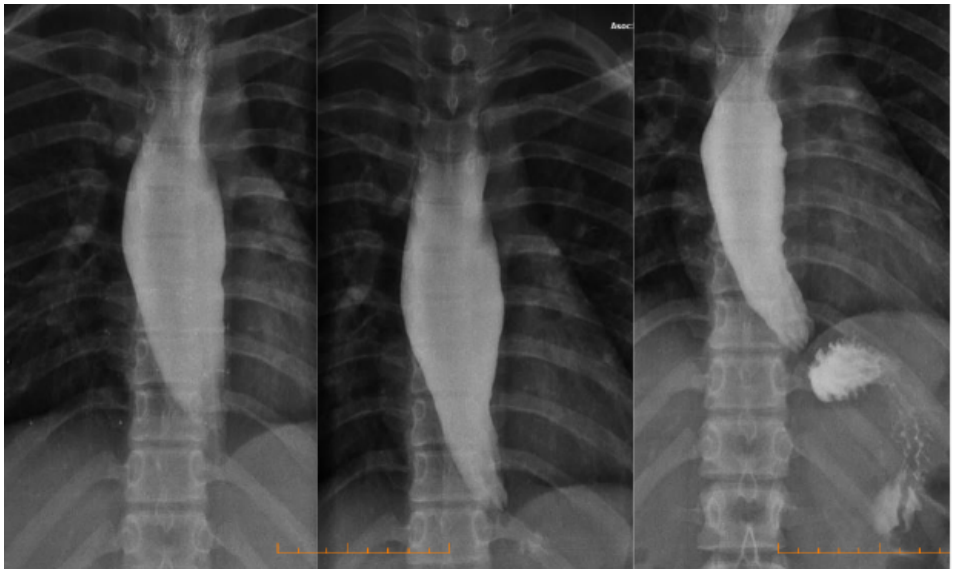
She had 2 vasovagal syncope episodes so she was admitted to our hospital suspected of a primary eating disorder.

On physical examination, she had decreased body mass, consistent with her weight loss, as the only positive sign. Her initial laboratory tests and abdominal ultrasound were normal.

During hospitalization, she was assessed by the mental health team, who ruled out a primary eating disorder.

The fact that the patient referred having dysphagia, accompanied by postprandial retrosternal pressure and regurgitation after consuming liquids or solids, strongly motivated the suspicion of achalasia. An upper gastrointestinal series found dilatation of the esophagus with filiform passage of contrast into the stomach and the presence of tertiary waves, compatible with achalasia (*Figure 1*). To complete her assessment, she underwent a high-resolution esophageal manometry, with 36 circumferential sensors and 12 impedance sensors (Medical Measurements System®), which confirmed the

Figure 1. Upper gastrointestinal series



Dilatation of the esophagus with filiform passage of contrast into the stomach.

diagnosis of type II achalasia. The serological tests for Chagas disease were negative.

She was managed with progressive esophageal balloon dilatation up to 15 mm, with adequate passage in the intraoperative control esophagography. At 72 hours, she had regurgitation again after starting a soft diet and an esophagography showed esophageal stricture. Given that dilatation failed, she underwent a laparoscopic Heller extramucosal myotomy with Dor fundoplication.

At 4 months, she progressively restarted with dysphagia and regurgitation, requiring a new esophageal balloon dilatation up to 20 mm. In the following 10 months, her clinical condition improved, her symptoms resolved, and her nutritional status was better.

DISCUSSION

Achalasia is an infrequent disease in pediatrics, with an incidence of 0.18–1.6 cases per 100 000 children/year and an age at diagnosis between 7 and 15 years.[1,3,4–10] There is no distinction in terms of race or sex,[3,4,5,9] although some publications have observed a certain predominance in males.[1,7,8]

It is an esophageal motility disorder resulting from the absence of inhibitory cells (adrenergic, nitric oxide-producing, and vasoactive peptide-producing cells) in the distal esophagus and lower esophageal sphincter (LES) of unknown etiology. This leads to lack of LES relaxation and alterations in normal esophageal peristalsis. Excitatory cholinergic activity is not counteracted by adrenergic inhibitory activity.[1–3,5,9] In most cases, it is an idiopathic condition.[1–5]

Secondary achalasia is associated with conditions that cause motor abnormalities similar to primary achalasia, such as amyloidosis, sarcoidosis, neurofibromatosis, eosinophilic esophagitis, multiple endocrine neoplasia syndrome type 2B, juvenile Sjögren syndrome, Fabry disease, and chronic idiopathic intestinal pseudo-obstruction.[4] The most frequent infectious cause is Chagas disease, although with the frequency of gastrointestinal involvement is low.[1,9] In our patient, based on clinical features and supplementary tests, these pathologies were ruled out.

The onset of signs and symptoms of achalasia is usually insidious, with progressive dysphagia, regurgitation, vomiting, postprandial retrosternal pain, and weight loss.[1,3,5,7] As in the case described here, the most frequent symptom leading to consultation is dysphagia, followed by regurgitation and weight loss.[1,7,8]

It is usually diagnosed late due to its low frequency and low initial index of suspicion.[1,3,6,7] It usually takes 4 to 6 months from the onset of symptoms to medical consultation.[1,3] Many pediatric patients are even referred to other specialists rather than to the gastroenterology department, mostly mental health services with a diagnosis of eating disorder, as was the case with our patient.[1,3] Achalasia should be considered in patients with suspected eating disorder who do not show the typical psychological profile. Some behaviors are similar, such as food selectivity or restriction, eating in a ritualistic manner (eating slow, chewing well, ruminating, or even mashing food), avoiding social situations that include eating, and exhibiting high levels of stress, depression, or anxiety. Other features of achalasia are distinctive, such as dysphagia, hunger, and desire to gain weight, regurgitation of undigested food, self-induced vomiting referred to relieve retrosternal pain and, mainly, the absence of body image disturbances. In most cases, the diagnostic delay is due to medical misinterpretation rather than an atypical presentation of achalasia.[3]

In case of clinical suspicion, a chest X-ray may find a widened mediastinum due to esophageal dilatation, even with hydro-aerial level inside and absence of gastric bubble.[8] An upper gastrointestinal series is the most accessible test, with a sensitivity of 95%; it shows a dilated esophagus with a typical “bird’s beak” morphological narrowing and also allows to rule out extrinsic compressions or malformations.[1,5,7,8] A manometry confirms the diagnosis and allows classifying achalasia into 3 subtypes according to the Chicago classification, when using a high-resolution manometry (HRM), which analyzes esophageal pressure topography, providing more information than a conventional linear manometry. Such distinction is relevant for treatment response and long-term prognosis. According to the pattern of intraluminal pressurization throughout the esophagus (pan-pressurization) and the type of contractions, 3 subtypes are established. Type I shows an absence of peristalsis (aperistalsis) and pan-pressurization is absent or less than 30 mmHg. Type II, the most frequent type, shows aperistalsis and pan-pressurization is greater than 30 mmHg. Type III, which is infrequent, shows spastic contractions, with or without periods of pan-pressurization in the esophageal topography.[1–5,7,9,10]

There are different therapeutic options,

although none of them are curative:[1,2,4–10]

An endoscopic injection of botulinum toxin decreases LES pressure; its effectiveness is comparable to surgery in the short term, but symptoms reappear after 6 to 12 months. Other drugs, such as nitrates or nifedipine, are even less effective and have greater adverse effects.[1,5,7–9]

An endoscopic pneumatic dilatation is effective, especially for type II achalasia.[4,8–10] Between 50% and 100% of patients require repeat procedures due to post-dilatation dysphagia, and between 30% and 75% require a myotomy due to long-term recurrence.[1,6,7]

In an extramucosal myotomy, the LES is weakened by sectioning muscle fibers. It increases the risk of post-operative gastroesophageal reflux (GER), so it is usually accompanied with anterior Dor fundoplication. It consists of wrapping the gastric fundus partially 180º around the LES, creating a reinforcement that protects the exposed mucosa, which prevents dissection of the posterior esophagus and has a lower reported incidence of post-operative dysphagia.[1,5–8] Some authors propose an anterior Dor fundoplication as the first therapeutic option because it has more lasting results and is minimally invasive via laparoscopy, with few differences among the different types of achalasia compared to endoscopic dilatation.[1,5,8–10]

An alternative surgical technique is peroral endoscopic myotomy, which has shown good results in limited pediatric series, but without studies comparing it with Heller myotomy in the long term.[1,2,4,6,7,9,10]

The case described here is characteristic of achalasia and, although balloon dilatation has been described as a good therapeutic option for type II achalasia, it was ineffective in our patient, who required a myotomy and subsequent dilatation.

It is important to know the clinical characteristics of this disease in order to achieve a timely diagnosis and effective treatment. The need for long-term follow-up is emphasized, due to the frequency of recurrence and the need for repeat procedures. ■

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