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CASE REPORT

# Idiopathic esophageal achalasia in a young patient: a case report

*Acalasia de esôfago idiopática em paciente jovem: um relato de caso*

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## ABSTRACT

**Introduction:** Esophageal achalasia is a condition characterized by impaired esophageal motility, defined by incomplete or absent relaxation of the lower esophageal sphincter (LES) associated with a lack of peristalsis in the esophageal body. It is a rare disorder that significantly impacts the patient's quality of life and nutritional status due to the presence of symptoms such as dysphagia, heartburn, regurgitation, weight loss, and other manifestations. **Methods:** Patient records were reviewed, and an extensive literature search was conducted to investigate the prevalence, etiology, pathophysiology, clinical manifestations, diagnosis, and treatment of achalasia, while correlating these findings with the presented case. **Case Report:** A 22-year-old female patient presented with a 6-month history of progressively worsening dysphagia, initially for solids and subsequently for liquids. This was accompanied by regurgitation, night cough, and an 8-kilogram weight loss within the period. Following upper gastrointestinal endoscopy, esophagram, and high-resolution esophageal manometry, a diagnosis of esophageal achalasia was made. Surgical treatment was chosen, resulting in clinical improvement and symptom resolution. **Conclusion:** This case emphasizes the importance of recognizing, diagnosing, and understanding the pathophysiology of achalasia to facilitate appropriate investigation and treatment, given its significant morbidity.

Keywords: Esophageal achalasia; Dysphagia; Esophagus.

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## RESUMO

**Introdução:** A acalasia de esôfago é uma doença caracterizada pelo comprometimento da motilidade esofágica, sendo definida pelo relaxamento incompleto ou ausente do esfíncter esofágico inferior (EEI) associado à aperistalse do corpo esofágico. É um distúrbio raro, responsável pelo comprometimento da qualidade de vida do paciente e do estado nutricional, uma vez que se apresenta com disfagia, pirose, regurgitação, perda de peso e outros sintomas. **Métodos:** Realizada revisão de prontuário do paciente, bem como extensa revisão bibliográfica sobre o tema, com estudo de prevalência, etiologia, fisiopatologia, manifestações clínicas, diagnóstico e tratamento, correlacionando aos achados da literatura com o caso apresentado. **Relato de Caso:** Paciente de 22 anos, sexo feminino, com quadro de disfagia há 6 meses, progressiva, inicialmente para sólidos e posteriormente para líquidos, associada à regurgitação, tosse noturna e perda ponderal de 8 quilogramas no período. Após realização de endoscopia digestiva alta, seriografia de esôfago e manometria esofágica de alta resolução foi realizado diagnóstico de acalasia de esôfago. Optou-se por tratamento cirúrgico, com melhora clínica e resolução dos sintomas. **Conclusão:** Ressalta-se, neste caso, a importância do reconhecimento, diagnóstico e compreensão da fisiopatologia da acalasia para que, dessa forma, sejam realizados o tratamento e a investigação adequados, visto sua alta morbidade.

**Palavras-chave:** Acalasia esofágica; Disfagia; Esôfago.

## Introduction

Esophageal achalasia is a pathology characterized by impaired esophageal motility, being defined by incomplete or decreased relaxation of the lower esophageal sphincter (LES) associated with aperistalsis of the esophageal. Clinically, patients report progressive dysphagia, heartburn, regurgitation, chest pain, and respiratory symptoms such as night cough. It is a rare disorder that affects both men and women equally, and often its diagnosis happens between the ages of 40 and 60. The etiopathogenesis is not yet scientifically clear, but there is an impairment of peristalsis and relaxation of the LES due to selective loss of inhibitory neurons of the myenteric plexus[1]. Some factors can serve as an initial trigger, such as neurotropic viruses or toxins, which will trigger an immune response in genetically predisposed individuals and cause chronic inflammation and neuronal loss[2].

## Case Report

A 22-year-old female patient from Igarapava-MG, born in Uberaba-MG and living in a brick house. Reports having a diagnosis of Attention Deficit Hyperactivity Disorder (ADHD) in the use of methylphenidate hydrochloride, without other comorbidities. In August 2022, began with progressive dysphagia, initially for solids evolving to liquids in 2 months, associated with immediate regurgitation after food intake. In addition, she reported frequent eructations, nocturnal cough, choking and weight loss of 8kg in the last 6 months.

In December 2022, the patient sought medical attention and treatment was initiated for eosinophilic esophagitis and lactose intolerance, with no clinical improvement. After 6 months she began treatment with the digestive surgery team, being requested a contrasted study of esophagus-stomach-duodenum (Figure 1), serology for Chagas disease, with non-reactive results, esophageal manometry (Figure 2) and upper digestive endoscopy (Figure 3). Surgical treatment was chosen, after the diagnosis of idiopathic achalasia, and esophagectomy was performed with cervical esophagogastric anastomosis, end-lateral with gastric fundus and surgical piece (Figure 4) was referred to anatomopathology (Figure 5). Patient was seen in Rheumatology for investigation of Achalasia, but no criteria of autoimmune disease were found.

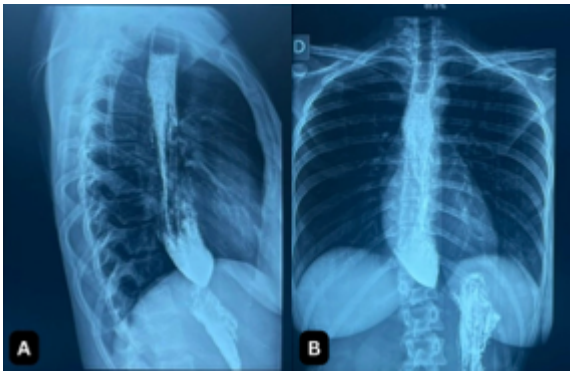

Figure 1. Contrasting X-ray of esophagus-stomach-duodenum performed on 01/10/2023. Moderate dilatation of the esophagus, distal tapering and slow emptying of the contrast into the stomach. Compatible with megaesophagus type III.

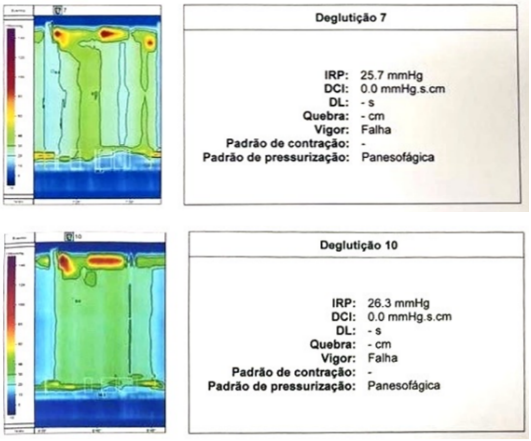


Figure 2. High resolution esophageal manometry performed on 01/26/2023 demonstrating type II achalasia. Dysfunction of the lower esophageal sphincter documented by the absence of relaxation, aperistalsis of the esophageal body with 100% of failure waves and 70% of swallows with panesophageal pressurization pattern.

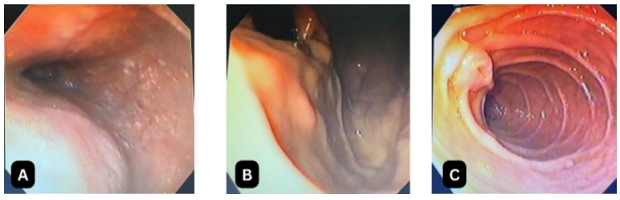

Figure 3. Upper gastrointestinal endoscopy performed on 06/12/2022. In A- Esophagus, with the presence of abundant fluids and semisolid remains retained in its cavity, progressing to the stomach, visualized during the examination. Impressive increase in the caliber of the distal esophageal Lumen, mucous membrane of blunt appearance. Mucosal changes coincide with the onset of gastric folds. Diaphragmatic hiatus 38 cm from ADS. In B- Entire gastric mucosa, absence of lesions in cardia, body and antrum. In C- Endoscope progresses up to second duodenal portion without other findings.

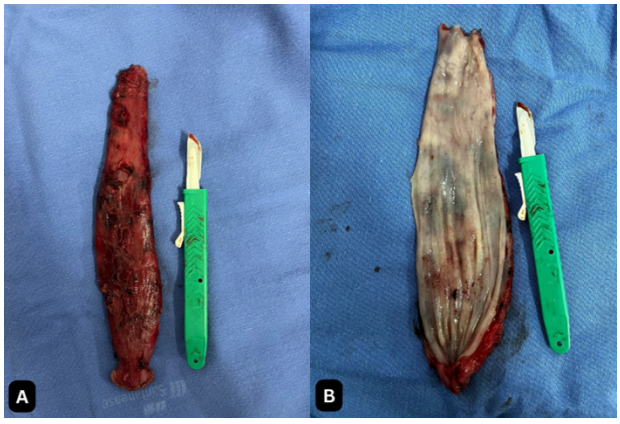

Figure 4. Segment of esophagus resulting from esophagectomy with 14.0 cm long. A. Ragged, reddish outer surface; B. Brown and soft mucous and a decrease in pleating. There is a hypertrophy of the muscle layer in the lower half. There are vinous dots on the mucosa of the dilated region, diffusely distributed.

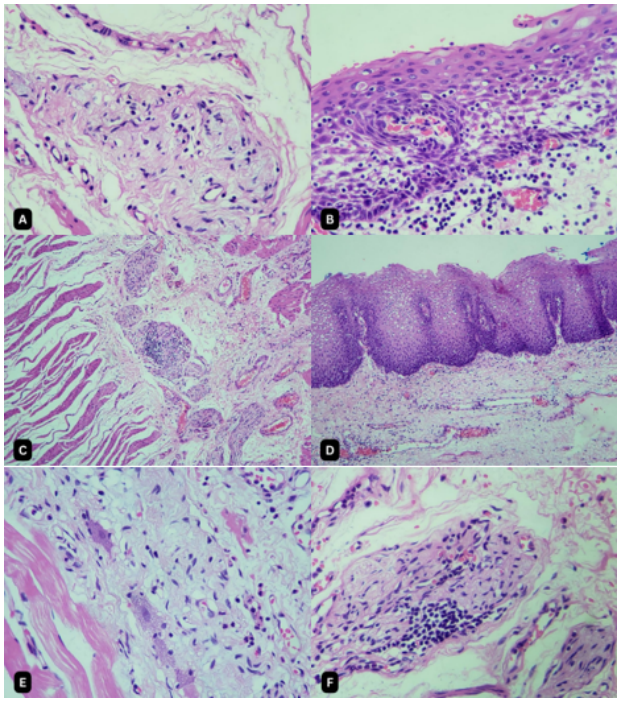

Figure 5. Anatomopathological result of esophagectomy product. **A.** Myenteric plexus nerve without viable ganglion cells, which exhibits chronic inflammatory process represented by lymphocytes (400x, HE); **B.** Squamous epithelium of the esophageal mucosa with reactive changes, edema, exocytosis of lymphocytes and congestion of papillae (400x, HE); **C.** Muscle layer with hypertrophy, myenteric nerve plexus with chronic inflammatory infiltrate (100x, HE); **D.** Esophageal Mucosa exhibiting superficial epithelium with acanthosis and submucosa with discrete edema and congestion (100x, HE); **E.** Myenteric nerve plexus with rare viable ganglion cells, with inflammatory infiltrate of lymphocytes and sparse neutrophils; **F.** Myenteric plexus nerve without viable ganglion cells, which exhibits chronic inflammatory process represented by lymphocytes (400x, HE).

## Discussion

Achalasia was first described in 1674 by Thomas Williams when he observed a pathology that occurred with a stop of the food bolus inside the esophagus for an unknown cause. The technological advance of diagnostic measures has stimulated interest in knowing about the etiology and pathophysiology of the disease[3,4].

It is a rare disorder of esophageal motility and epidemiological data are scarce, with some studies indicating an incidence of 0.03 to 1.63 per 100.000 people per year and a prevalence of 10 cases per 100.000 individuals. It is estimated that approximately 27-42% of patients with achalasia used to be misdiagnosed because of their overlapping symptoms such as heartburn and chest pain. A recent American epidemiological survey conducted in areas with high availability of high - resolution manometry found that the annual prevalence and incidence of Achalasia were 162.1/100.000 and 26.0/100.000, respectively, being at least 2-3 times higher than previously estimated. Therefore, given the poor distribution of the most advanced diagnostic criteria and technologies, including esophageal manometry, it is safe to say that the actual number of patients with achalasia worldwide remains unknown[2,5-7].

Although it can occur in any age group, the diagnosis is usually made between the fourth and sixth decade, affecting men and women equally and without ethnic predilection. It is a chronic disease that affects the morbidity and quality of life of patients and, in general, takes time to be correctly diagnosed[2,5,6].

Achalasia can be divided into primary (idiopathic) or secondary. Primary is the most common in the world population, with the exception of the South American region. Its etiology is still debated despite established hypotheses. Secondary achalasia is more common in South American countries and has its etiology known. Most cases are associated with degeneration of myenteric plexuses in Chagas disease, infection caused by protozoa *Trypanosoma Cruzi*, endemic to the region. There was many actions against the vector of Chagas disease, decreasing the prevalence of the pathology in the country and, thus, most of the cases currently observed are chronic. Due to the patient's symptomatology and the diagnostic hypothesis of Achalasia secondary to Chagas disease, serology was requested, being non-reactive[3,8,9].

The etiopathogenesis of idiopathic achalasia is related to autoimmunity, which triggers inflammatory processes responsible for the degradation of inhibitory innervation of the myenteric plexus and LES fibers, leading to an imbalance between excitatory and inhibitory activity. Acetylcholine-mediated excitatory activity unopposed by inhibitory activity performed by nitrergic neurotransmitters and vasoactive intestinal peptide cause sphincter relaxation failure and aperistalsis. Viral infection, idiopathic autoimmune triggers and genetic predisposition are some mechanisms that can serve as a factor to trigger the chronic inflammatory process and lead to achalasia. Related genes include genes linked to the immune system and neurodegeneration[5,7,10].

The viral infections most associated with this pathology are the viruses of the herpes family, paramyxovirus and HIV. In contrast, not all patients with these viral infections develop achalasia, demonstrating that chronic infection associated with individual genetic and environmental factors can facilitate neuronal loss. The autoimmune etiology is related to the presence of T cells and eosinophils in the myenteric plexus, anti-myenteric antibodies and increased prevalence of human leukocyte antigens (HLA) class II, and may be related to other autoimmune diseases, such as uveitis, Sjogren's Syndrome, lupus erythematosus, type 1 diabetes, hypothyroidism, rheumatoid arthritis, scleroderma, ankylosing spondylitis, myasthenia gravis, psoriasis and others. These pathologies have a higher prevalence in the population with achalasia in the younger age group. Thus, the existence of familial cases of Achalasia suggests that genetics may be a predisposing component to the development of the disease. The patient in this case did not present clinical symptoms of current viral infection, nor did she meet sufficient criteria for the diagnosis of autoimmune disease, and reevaluation was requested within one year by Rheumatology[11,12].

Dysphagia is the main symptom observed in patients with achalasia, which is initially caused by solid foods, but evolves to liquids in most cases. It may be associated with regurgitation of undigested food or saliva. Weight loss is a common find when the diagnosis is made. Dysphagia, regurgitation and weight loss make up the classic triad of Achalasia. Chest pain may be part of the symptomatology, and angina of cardiac origin should be excluded. Heartburn may appear in some individuals. In addition, respiratory complications are the most common to occur, such as aspiration pneumonia and lung abscess. It is important to investigate achalasia in patients who have dysphagia and regurgitation who do not respond to treatment with proton pump inhibitors[13-15].

The upper digestive endoscopy is the initial examination to investigate symptoms of the gastrointestinal tract. Although it is not the gold standard for the diagnosis of motor disorders, upper digestive endoscopy is important to exclude structural or mucosal changes in the esophagus and Cardia, such as peptic stenosis, cancer and eosinophilic esophagitis, situations where there is dysphagia (pseudoacalasia). Esophageal manometry (Figure 2) is the gold standard test for the diagnosis of esophageal motility disorders. The manometric finding of aperistalsis and incomplete relaxation of the LES without evidence of mechanical obstruction corroborate the diagnosis of Achalasia. The barium esophagogram, as shown in Figure 1, can also be used in the propaedeutics of suspected cases of Achalasia, demonstrating esophageal dilatation, narrowing of the esophagogastric junction (aspect of "parrot beak"), aperistalsis and deficiency in barium emptying[1,5].

In the aperistaltic esophagus there is greater contact of irritants and carcinoegenes epithelial lining due to stasis.

And as the desease progresses, especially after 5 to 15 year, there is a greater risk of cancer. This risk is higher for squamous cell carcinoma and adenocarcinoma compared to the general population and more prevalent in males[2,16].

Depending on the severity of the disease the treatment is instituited. Pneumatic balloon dilation is one of the first recommendations, having good long-term results. Oral pharmacotherapy and botulinum toxin should be considered for patients with clinical conditions that do not favor endoscopic or surgical treatment or with therapeutic failure after these procedures. Among the most studied drugs for cases of Achalasia, beta-agonists, calcium channel blockers, anticholinergics and nitrates can be mentioned, being recommended for patients with early-stage disease or as a temporary measure before another approach. Endoscopic treatment can be carried out using sclerosing agents and ***stents***. Surgical treatment has changed dramatically since the introduction of minimally invasive surgeries and can be instituted through cardiectomy, cardioplasty, cardiomyotomy or esophagectomy. These procedures should be considered in advanced cases of Achalasia. In the reported case, the patient was informed about the available therapeutic options and, in consensus with the Digestive System Surgery team, it was decided to perform an esophagectomy, aiming to meet the patient's expectations[1,5,17-20].

## Conclusion

Achalasia is a condition characterized by impairment of the lower esophageal sphincter and peristalsis of the esophageal body. It can be primary (idiopathic) or secondary, mainly due to Chagas disease. The etiopathogenesis of idiopathic achalasia is related to autoimmunity, also viral infections, idiopathic autoimmune trigger, and genetic predisposition may be involved in this process. The classic symptoms consist in progressive dysphagia, regurgitation, and weight loss. Esophageal manometry is the gold standard for diagnosis, but upper gastrointestinal endoscopy is the initial examination for investigating cases. Treatment should be tailored according to disease severity. Oral pharmacotherapy and endoscopic treatment are possible options.

It is concluded that much still needs to be studied about this condition, regarding etiology, evolution and the best way to follow up these patients, since, to date, the guidelines of the American Society of Gastrointestinal Endoscopy (ASGE) and the guidelines of the American College of Gastroenterology (ACG) 2020 cite the lack of sufficient evidence to recommend endoscopy as a routine screening in the Prevention of cancer as a complication[2].

## Author's Contribution

We describe contributions to the papers using the taxonomy (CRediT) provided below: *Conceptualization, Investigation, Methodology, Visualization & Writing – review & editing:* Gabriel Santos Faria de Carvalho, Ana Júlia Maluf Coelho. *Project administration, Supervision & Writing – original draft:* Gabriel Santos Faria de Carvalho, Ana Júlia Maluf Coelho, Mariana Almeida Hein, José Miguel da Silva Maciel, Tullio Novaes Silva, Ana Flávia Carrijo Chiovato, Silvia Maria Perrone Camilo. *Validation & Software:* Gabriel Santos Faria de Carvalho, Ana Júlia Maluf Coelho, Ana Flávia Carrijo Chiovato, Silvia Maria Perrone Camilo. *Resources & Funding acquisition:* Ana Flávia Carrijo Chiovato, Silvia Maria Perrone Camilo. *Data curation & Formal Analysis:* Ana Flávia Carrijo Chiovato, Silvia Maria Perrone Camilo.

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