

Ten Tight Years: A Late Eosinophilic Esophagitis Diagnosis

Dez Anos de Aperto: Um Diagnóstico Tardio de Esofagite Eosinofílica

Pedro Sonié^{1*}, Sara Teixeira¹, Marta Perro Neves¹, Ana Rita Duarte¹,
Maria Elvira Pinto¹

*Autor Correspondente/Corresponding Author: Pedro Sonié

Pedro Sonié [pedrosonie11@gmail.com]

Rua Barão do Corvo nº 676, 4400-037 Vila Nova de Gaia, Portugal

ORCID iD: 0000-0002-1651-4351

RESUMO

A esofagite eosinofílica é atualmente um dos diagnósticos mais frequentes na avaliação de dificuldade alimentar em crianças e disfagia e impactação alimentar em adultos. O nosso caso clínico reporta um homem de 20 anos, com antecedentes de asma, que desenvolveu disfagia para sólidos e impactação alimentar ao longo de um período de 10 anos. O diagnóstico tardio levou ao desenvolvimento de sintomas incapacitantes, achados endoscópicos avançados e as opções terapêuticas com maiores riscos associados. Os médicos de família têm um papel essencial no diagnóstico correto e atempado destes sintomas e referência apropriada para a consulta da especialidade.

PALAVRAS-CHAVE: Cuidados de Saúde Primários; Esofagite Eosinofílica/diagnóstico; Perturbações da Deglutição

1. USF Arco do Prado, ACeS Grande Porto VII, Gaia, Portugal.

Recebido/Received: 2022/04/10 - Aceite/Accepted: 2023/08/28 - Publicado online/Published online: 2023/09/15 - Publicado/Published: 2023/09/30

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ABSTRACT

Eosinophilic esophagitis is now one of the most common conditions diagnosed during the assessment of feeding problems in children and during the evaluation of dysphagia and food impaction in adults. We report a case of a 20-year-old male, with a history of asthma, who developed solid dysphagia and food impaction symptoms over a 10-year course. A late diagnosis led to incapacitating symptoms, advanced endoscopic findings, and riskier therapeutic options. Primary care providers are pivotal for timely and accurate detection of symptoms and proper referral to specialists.

KEYWORDS: Deglutition Disorders; Eosinophilic Esophagitis/diagnosis; Primary Health Care

INTRODUCTION

Eosinophilic esophagitis is now one of the most common conditions diagnosed during the assessment of feeding problems in children and during the evaluation of dysphagia and food impaction in adults. The entity exists worldwide but has been most extensively studied in Western countries, where its prevalence has been estimated to be 0.4% among all children and adults.¹ Diagnosis requires greater than or equal to 15 eosinophils per high-power field on light microscopy.² Primary care providers are pivotal for timely and accurate recognition of symptoms of eosinophilic esophagitis, for facilitating diagnoses through specialist referrals, and for understanding management strategies.³ We report a case of a 20-year-old male who was diagnosed with eosinophilic esophagitis 10 years after symptom onset.

CASE REPORT

A 20-year-old male, Caucasian, with a history of asthma, and as needed salbutamol as only regular medication. Had an irregular follow-up at his primary health care unit and was followed by a pediatrician. Back in 2012, at an appointment in his health care unit was described as having a good physical development, academic achievement, and socialization. No major findings at physical examination and had a body mass index of 17.4 kg/m². After this appointment he presented two times at his health care unit for acute gastrointestinal and osteoarticular symptoms in 2014. In 2017 he had a surveillance appointment where he first talked about these complaints. An upper endoscopy and a thyroid ultrasound were prescribed, which were never made by the patient.

In January 2022, he presented to his primary health care doctor with solid dysphagia, and food impaction. Occasionally he had to provoke the vomit to alleviate the symptoms. He reported no heartburn, chest pain or dysphonia. These symptoms developed progressively over a period of 10 years. He managed to adapt to his

complaints by eating slowly and with extensive chewing. Reported severe impact in his social activities, with inhibition to participate in social events where a meal was mandatory. Once again physical examination revealed no major findings, and the body mass index was 23.0 kg/m².

At this appointment an upper endoscopy was prescribed which revealed “trachealization of esophagus, esophageal rings and at 25 cm an unsurpassable stenosis”. Epithelium biopsy for histopathological evaluation was performed and revealed “squamous epithelium with basal cell hyperplasia, spongiosis and permeation by polymorphic inflammatory cells, rich in eosinophils. More than 15 eosinophils per high-powered microscopic field. No eosinophilic microabscess found”. The patient was then urgently referred to a gastroenterology appointment.

DISCUSSION

The diagnosis of eosinophilic esophagitis is based upon symptoms, endoscopic appearance, and histological findings. Eosinophilic esophagitis (EoE) should be suspected in adolescents and adults when the predominant complaint is esophageal dysphagia,⁴ which is reported by 60% to 100% of patients,⁴⁻⁶ and food impaction can be seen in more than 25%.⁷ Heartburn (30%-60%) and noncardiac chest pain (8%-44%) are commonly reported.^{4,7} Abdominal pain, nausea, vomiting, diarrhea, gastrointestinal bleeding, and weight loss are uncharacteristic in adults with EoE, and a different process or more diffuse eosinophilic gastrointestinal disorder (eg, eosinophilic gastroenteritis, eosinophilic colitis) should be considered when these features predominate.⁸

Since the symptoms of EoE are not specific, the diagnosis may be missed. In one retrospective study that included 200 patients with symptomatic EoE, the median delay in diagnosis was six years (interquartile range 2 to 12 years).⁹

There is a strong association of EoE with allergic conditions such as food allergies, environmental allergies, asthma, and atopic dermatitis. It has been estimated that 28% to 86% of adults and 42% to 93% of children with EoE have another allergic disease.^{10,11}

The possibility of disease progression was supported in a case-control study that suggested an increased rate of dysphagia (49% vs 6%) and food impaction (40% vs 3%) in children with esophageal eosinophilia who had been followed for an average of 15 years.⁴ Other studies show that the best strategies for preventing the onset of strictures appear to include treatment early in the disease course and long-term control of esophageal eosinophilic inflammation retention.¹²

The gold standard for EoE diagnosis remains biopsy findings demonstrating increased intraepithelial esophageal eosinophil counts without concomitant eosinophilic infiltration in the stomach or duodenum. At least 5 biopsy specimens should be obtained at multiple esophageal levels to maximize the sensitivity based on a diagnostic threshold of 15 or more eosinophils per high-power field.¹³

Therapies include proton pump inhibitors, elimination diets, and topical corticosteroids. Effective treatment can reverse tissue fibrosis in some patients, as well as decrease the rate of food impactions. Esophageal stricture or narrowing is treated best by dilation, which is safe and effective when done cautiously. This is an important treatment to improve symptoms of dysphagia, but it does not impact the underlying eosinophilic inflammation.¹⁴

We report a case in which the patient revealed typical symptoms for about ten years, and had a history of asthma, which should have raised the suspicion for this disease and prompt the clinical investigation. As previously referred, there is evidence for disease progression, and therefore an earlier diagnosis could have spared the need for dilation. A meta-analysis determined a 0.3% perforation risk for this technique.¹⁵ Also it could have spared years of debilitating symptoms and social inhibition through the patient's adolescence.

This case report aims to raise awareness that primary care providers are pivotal for timely and accurate detection of symptoms potentially related to EoE, referral to proper specialists for diagnosis, coordination of care between multiple providers, as well as transition of care from pediatric to adult providers, all with the goal of improving patient quality of life and reducing long-term EoE complications.³

DECLARAÇÃO DE CONTRIBUIÇÃO/ CONTRIBUTORSHIP STATEMENT:

PS, ST, MN, AD e MP: Conceção, escrita, revisão do artigo e aprovação da versão final

PS, ST, MN, AD and MP: Design, writing, article review and approval of the final version

RESPONSABILIDADES ÉTICAS

CONFLITOS DE INTERESSE: Os autores declaram a inexistência de conflitos de interesse na realização do presente trabalho.

FONTES DE FINANCIAMENTO: Não existiram fontes externas de financiamento para a realização deste artigo.

CONFIDENCIALIDADE DOS DADOS: Os autores declaram ter seguido os protocolos da sua instituição acerca da publicação dos dados de doentes.

CONSENTIMENTO: Consentimento do doente para publicação obtido.

PROVENIÊNCIA E REVISÃO POR PARES: Não comissionado; revisão externa por pares.

ETHICAL DISCLOSURES

CONFLICTS OF INTEREST: The authors have no conflicts of interest to declare.

FINANCING SUPPORT: This work has not received any contribution, grant or scholarship.

CONFIDENTIALITY OF DATA: The authors declare that they have followed the protocols of their work center on the publication of data from patients.

PATIENT CONSENT: Consent for publication was obtained.

PROVENANCE AND PEER REVIEW: Not commissioned; externally peer reviewed.

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