GE Port J Gastroenterol. 2014;21(5):203–207

Portuguese Journal of Gastroenterology
Jornal Português de Gastrenterologia
www.elsevier.pt/ge

CLINICAL CASE

An uncommon cause of dysphagia in pediatric age

Joana Aquino[a,*], **Patrícia Rocha**[a], **Susana Almeida**[b], **Alexandra Luz**[a]

[a] *Serviço de Pediatria do Centro Hospitalar de Leiria (CHL), Leiria, Portugal*
[b] *Unidade de Gastrenterologia do Hospital Pediátrico Carmona da Mota, Centro Hospitalar e Universitário de Coimbra, Coimbra, Portugal*

Received 10 April 2014; accepted 18 June 2014
Available online 14 August 2014

KEYWORDS
Achalasia;
Dysphagia;
Esophageal manometry;
Teenager

Abstract
Introduction: Achalasia is a rare disease in children of unknown etiology. For its rarity and diagnostic difficulty, the authors report the case of a teenager with achalasia.
Case report: 15-year-old boy, with unremarkable past medical history, was referred to the outpatient clinic with a 3-month history of regurgitation and dysphagia. An upper digestive endoscopy was performed, which was normal. The symptoms got worse and he lost 9% of weight. Considering the diagnosis of eating behavior disorder, he was admitted for further investigation. Laboratorial evaluation was unremarkable. Dysphagia characterization suggested a disorder of esophageal motility. Barium follow-through was compatible with achalasia and high-resolution esophageal manometry confirmed this diagnosis. He underwent laparoscopic Heller myotomy combined with Dor fundoplication with no symptom recurrence.
Comments: Achalasia is a rare disease associated with a challenging and delayed diagnosis. The normality of the upper digestive endoscopy and the hypothesis of an eating behavior disorder both led to a delayed diagnosis. It is important to proceed with investigation in the presence of unremitting dysphagia symptoms.

PALAVRAS-CHAVE
Acalásia;
Disfagia;
Manometria;
Adolescente

Uma causa de disfagia pouco comum em idade pediátrica

Resumo
Introdução: A acalásia é uma doença rara em idade pediátrica, de etiologia desconhecida. Pela sua raridade e dificuldade diagnóstica, os autores relatam o caso de um adolescente com acalásia.

* Corresponding author.
E-mail address: joanamgaquino@gmail.com (J. Aquino).

http://dx.doi.org/10.1016/j.jpg.2014.06.001

Caso clínico: Adolescente de 15 anos, sexo masculino, com antecedentes pessoais irrelevantes. Por queixas de regurgitação e disfagia com 3 meses de evolução, efetuou Endoscopia Digestiva Alta (EDA) que foi normal. Após agravamento das queixas, com perda ponderal (9%), foi colocada a hipótese diagnóstica de perturbação do comportamento alimentar, pelo que foi internado para esclarecimento do quadro. Analiticamente não apresentava alterações. As características da disfagia durante o internamento (inicialmente para líquidos e posteriormente também para sólidos) sugeriram alteração da motilidade esofágica, tendo sido realizado trânsito esofágico baritado, cujo resultado foi compatível com a hipótese de acalásia. A manometria esofágica de alta resolução confirmou este diagnóstico. Foi submetido a miotomia laparoscópica de Heller com fundoplicatura de Dor, sem recorrência das queixas.
Comentários: A acalásia é uma doença rara, associada a dificuldade e atraso no diagnóstico. A normalidade da EDA e a hipótese de perturbação do comportamento alimentar, contribuíram para atrasar o diagnóstico, pelo que, na presença de queixas persistentes de regurgitação ou disfagia é importante caracterizar exaustivamente os sintomas, pensar na doença e prosseguir com a investigação.

Introduction

Achalasia is a primary motor disorder of the esophagus characterized by insufficient relaxation of the lower esophageal sphincter (LES) and an absence of peristalsis in the esophageal body.[1,2] Its clinical manifestations are difficulty in the passage of food through the esophagogastric junction, without a true organic stenosis or an extrinsic compression, as in the case of a functional esophageal obstruction.[1–3] It leads to incomplete emptying of the esophagus, with a gradually esophageal dilatation and its consequences.[4]

The disease was first described by Thomas Willis in 1674 and, in 1953, King described the first case of achalasia in childhood, presenting the story of a 6-month-old infant.[2,4–6]

This is a rare condition, especially in the pediatric age range,[2] being more frequent in adults between 25 and 60 years old.[5] It is a disease with an incidence of 1:100,000 in the general population, with only 4–5% of these cases occurring in children.[3–7,12] Few patients present symptoms before 15 years of age.[6] In children, the average age at diagnosis is 8.8 years.[8]

The influence of genetic factors in the etiology of achalasia is suggested by the existence of this disease associated with some syndromes, such as Allgrove Syndrome, Rozycki Syndrome and Pierre–Robin Syndrome[3]. Reports of isolated familial achalasia represent less than 1% of all cases of this disease.[3]

The etiology of idiopathic achalasia is unknown.[1,2] There are some forms of secondary achalasia, such as pseudoachalasia secondary to infiltration by tumor, and Chagas Disease which is associated with infection by *Trypanosoma cruzi*.[1,7,9]

Regarding its pathophysiology, achalasia is associated with functional loss of ganglion cells of the myenteric plexus of the esophagus. This change appears to be caused by an autoimmune response involving cytotoxic T-cells and autoantibodies, which triggers a process of ganglionitis with neuron loss of the esophageal myenteric plexus. This trigger can be of an infectious nature associated with some viral infections (Herpes Simplex Virus type 1, measles) or from the direct effect of a toxin, in a susceptible host (immunogenic base).[3,5,9,10] The inflammation of the myenteric plexus leads to degeneration and dysfunction of postganglionic inhibitory neurons of the distal esophagus. The imbalance between nitric oxid and vasoactive intestinal peptide used by neurons as neurotransmitters translates in a deregulation in excitatory and inhibitory control of the LES and the adjacent esophagus, leading to achalasia.[9,10]

Given the rarity of this disease, especially in children[7], the description of new cases may contribute to a better knowledge of its clinical manifestations and evolution.

Case report

Male adolescent, 15-year-old, with no relevant past familiar or personal medical history, was referred to the Outpatient Clinic of Centro Hospitalar de Leiria by his attending physician because of borderline elevated TSH (5.81 μUI/mL). This analysis followed complaints of sporadic regurgitation of undigested food which started 3 months earlier. He denied anorexia, abdominal pain, change in stools, weight loss or fever. Approximately 2 months later he complained of dysphagia, initially for liquids and subsequently also for solid foods. He then stated daily nocturnal regurgitation of undigested food.

At physical exam he weighed 53.5 kg (p25–p50); Height – 174 cm (p50–p75); Body Mass Index (BMI) – 17.7 kg/m^2 (p10–p25). He was longilineal with no palpable thyroid or cutaneous lesions. He was flushed and hydrated. Cardiopulmonary auscultation and abdominal exam found no alterations.

Laboratory tests showed: WBC 9500/μL; Neutrophils 4200/μL; Lymphocytes 4300/μL; Eosinophils 300/μL; Hemoglobin 15.0 g/dL; VS 1 mm in 1st hour; TSH 3.39 μUI/mL and FT4 10,6 pmol/L. Liver function and biochemistry with