Rev Paul Pediatr. 2015;33(3):371–375

REVISTA PAULISTA DE PEDIATRIA

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CASE REPORT

Pathologic aerophagia: a rare cause of chronic abdominal distension

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Received 11 November 2014; accepted 18 January 2015
Available online 30 June 2015

KEYWORDS
Aerophagy;
Pathologic dilatation;
Gastric emptying

Abstract
Objective: To describe an adolescent with pathologic aerophagia, a rare condition caused by excessive and inappropriate swallowing of air and to review its treatment and differential diagnoses.
Case description: An 11-year-old mentally impaired blind girl presenting serious behavior problems and severe developmental delay with abdominal distension from the last 8 months. Her past history included a Nissen fundoplication. Abdominal CT and abdominal radiographs showed diffuse gas distension of the small bowel and colon. Hirschsprung's disease was excluded. The distention was minimal at the moment the child awoke and maximal at evening, and persisted after control of constipation. Audible repetitive and frequent movements of air swallowing were observed. The diagnosis of pathologic aerophagia associated to obsessive–compulsive disorder and developmental delay was made, but pharmacological treatment was unsuccessful. The patient was submitted to an endoscopic gastrostomy, permanently opened and elevated relative to the stomach. The distention was resolved, while maintaining oral nutrition.
Comments: Pathologic aerophagia is a rare self-limiting condition in normal children exposed to high levels of stress and may be a persisting problem in children with psychiatric or neurologic disease. In this last group, the disease may cause serious complications. Pharmacological and behavioral treatments are ill-defined. Severe cases may demand surgical strategies, mainly decompressive gastrostomy.

DOI of original article: http://dx.doi.org/10.1016/j.rpped.2015.01.003
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PALAVRAS-CHAVE
Aerofagia;
Distensão patológica;
Esvaziamento gástrico

Aerofagia patológica: uma causa rara de distensão abdominal crônica

Resumo
Objetivo: Descrever o caso de uma adolescente com aerofagia patológica, uma doença rara causada pela deglutição excessiva e inapropriada de ar, e revisar o tratamento e os diagnósticos diferenciais.
Descrição do caso: Menina de 11 anos portadora de retardo mental e cegueira, apresentava problemas comportamentais associados a retardo do desenvolvimento, foi consultada por distensão abdominal persistente por oito meses. Sua história pregressa incluía uma fundoplicatura à Nissen. Tomografia e radiografias abdominais mostravam distensão difusa do trato digestivo por ar, incluindo cólon e delgado. Doença de Hirschsprung foi excluída. A distensão persistiu mesmo após o controle da constipação e era mínima de manhã e máxima à noite. Ruídos audíveis e repetitivos de deglutição de ar foram observados e auscultados. A criança foi tratada farmacologicamente com o diagnóstico de aerofagia patológica associado a distúrbio obsessivo compulsivo, sem sucesso. A paciente foi submetida a gastrostomia descompressiva endoscopia e manteve nutrição oral.
Comentários: A aerofagia patológica é uma doença rara e autolimitada em crianças, mas pode ser um problema grave e persistente naquelas com problemas neuropsiquiátricos, nas quais pode causar complicações sérias. Os tratamentos comportamentais e farmacológicos têm pouco sucesso nesse grupo. Casos graves podem precisar de tratamento cirúrgico, principalmente gastrostomia descompressiva.

Introduction

Pathologic aerophagia, caused by excessive and inappropriate swallowing of air, is a rare condition in clinical Pediatrics. The disease affects mainly normal children dealing with stress situations, but approximately a quarter present severe psychiatric and/or neurologic problems. Differential diagnosis primarily involves other gastrointestinal dysfunctional syndromes.

We describe herein a case of severe pathologic aerophagia in a school-aged mentally impaired child and review the literature on the theme.

Case description

An 11-year-old mentally impaired girl was transferred from another hospital to the Pediatric ward of Antonio Pedro University Hospital, Rio de Janeiro, Brazil, to be investigated for possible aganglionic megacolon. The girl was blind, presented serious behavior problems and severe developmental delay and was unable to cooperate with anamnesis and physical examination. Her mother recalled a history of abdominal distension only from the last 8 months, unresponsive to repetitive enemas and associated with pain crises that demanded repetitive consultations to Pediatric emergency services. Moderate constipation was noted since the girl was a toddler and dealt with by occasional usage of oral laxatives. Appetite was preserved during the crises and there were no signs of gastric dysfunction, retching or vomiting.

Her physical examination showed a seriously undernourished prepubertal girl —14.5 kg (<5th percentile of World Health Organization growth curve);[1] 102cm (<5th percentile of World Health Organization growth curve)[1], with severe diffuse abdominal distension (abdominal circumference 76cm at the level of the umbilicus) (Fig. 1). Peristalsis was normal. There were no palpable masses or abdominal pain. Anal and rectal examination demonstrated a normotonic topic anus. The rectum was full of solid feces.

Neuropsychiatric evaluation confirmed severe developmental delay and disproportionate aggressiveness toward the medical team. Several repetitive stereotyped move-

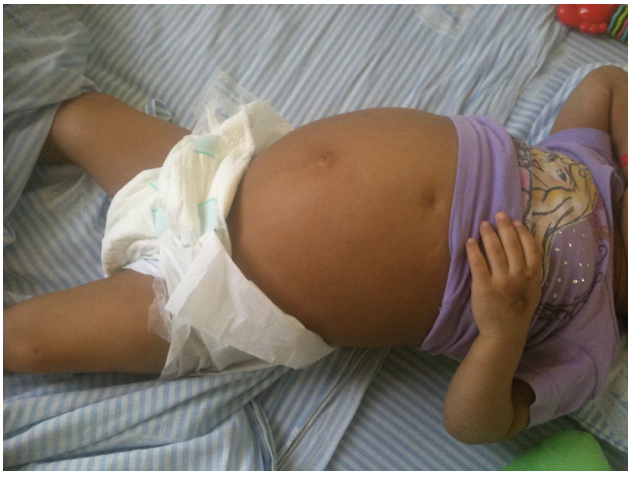

Figure 1 Severe abdominal distension, causing limitation of thoracic expansion. Abdominal scar corresponds to previous gastrostomy site. Notice scarring on the hand, due to repetitive biting.

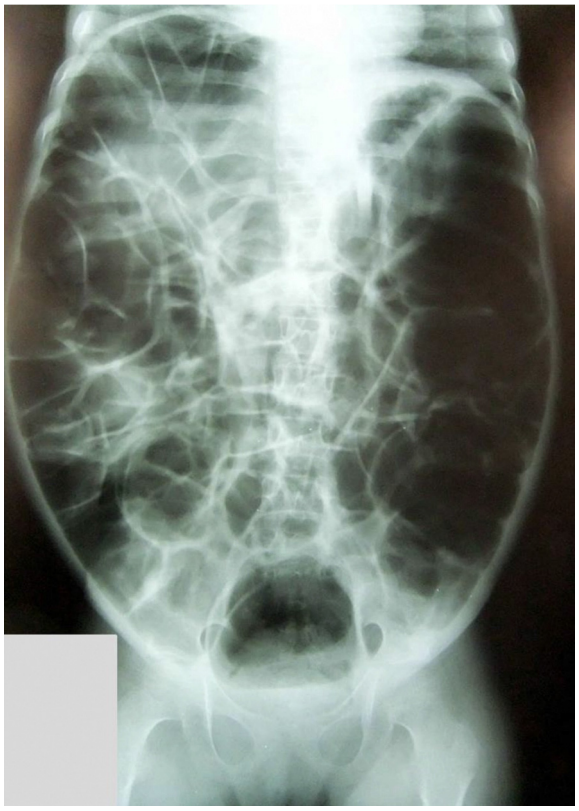

Figure 2 Diffuse and severe gas distension, attaining small intestine and colon.

ments (repetitive biting of the hands, moving the thorax forward and backwards and agitating a chatterbox) were observed. The patient was in chronic use of pericyazine (8 mg in the morning and 10 mg at night) that was gradually withdrawn during admission.

The child was born prematurely after a twin gestation. She presented congenital cataract and was submitted to a fundoplication and gastrostomy at 3 months old to treat swallowing problems, repetitive aspiration and symptomatic gastroesophageal reflux. After a difficult perinatal period the patient was abandoned by the mother and institutionalized. Her gastrostomy was taken off and she was then orally fed. The girl was adopted at 2-years-old, together with two other neurologically impaired children. The family already had other natural children and the father is wheelchair-bound.

An abdominal CT and several abdominal radiographs showed diffuse gas distension of the small bowel and colon (Fig. 2).

Her constipation was controlled with daily enemas for 15 days, after which the girl evacuated spontaneously daily. A rectal biopsy excluded Hirschsprung's disease. Clinical observation demonstrated that her distension was minimal at the moment the child awoke in the morning (mean abdominal circumference of 68cm) and maximal at evening (mean abdominal of circumference 74cm). Audible repetitive and frequent movements of air swallowing were observed. To test the hypothesis of pathologic aerophagia in a patient incapable of burping, her abdominal perimeter was measured (76cm) and a nasogastric catheter was installed and opened for 24 h. At the end of the period, her abdominal perimeter was 61cm. A few hours after taking off the catheter her abdominal perimeter reached 78cm.

Departing from the diagnosis of pathologic aerophagia associated to obsessive–compulsive disorder and developmental delay, treatment with risperidone 1.5mg/day (0.5mg in the morning and 1.0mg at night) for 3 weeks was unsuccessful. The usage of oral carbon was considered unpractical.

The child was submitted to an endoscopic gastrostomy without complications. The family was taught to maintain the gastrostomy opened and elevated >30cm relative to the stomach as frequently as possible, to allow the elimination of swallowed air, avoid the loss of gastric secretions and at the same time enable normal oral nutrition. This was possible by using an elongating device connected to the gastrostomy tube and fixed to the anterior thoracic wall in a superior direction when the child was sitting or standing, during the day, and the chronic abdominal distension resolved. The child, however, maintained the repetitive swallowing movements.

At the moment of this report, the family reports constant drainage of air from the gastrostomy and recurrence of the abdominal distension whenever the gastrostomy stays closed for a long period. Chronic constipation did not recur. The girl stays under psychotherapy treatment and social services supervision.

Discussion

In children, 70% of the bowel gas comes from swallowed air.[2] The radiological expression of aerophagia is the presence of gas throughout the small bowel, which is physiologic in newborns and infants, but limited to specific situations in older children.

Rome criteria defined pathological aerophagia as air swallowing causing abdominal distension and/or repetitive flatulence/belching, present for >12 weeks in a year. This standard may be problematic in mentally impaired children presenting constipation and previously submitted to fundoplication, both of which are common in this population. Those patients are frequently unable to burp and rectal fecal masses limit the elimination of flatus. Only our patient and two others reported cases[3,4] have been previously submitted to fundoplications, which may have exaggerated their pathological aerophagia, due to the impossibility of burping and belching. Most patients with pathological aerophagia are school-aged children and adolescents. The disease is rare and chronic: almost half of the patients present symptoms for $\geq$1 year at the time of diagnosis.

Psychiatric diseases attain 1/4 of pathological aerophagia cases, mostly mental retardation, autism and Rett syndrome.[5] Although pathological aerophagia is rare, 8.8% of institutionalized mentally impaired children present aerophagia.[2] Pathological aerophagia may represent the more severe spectrum of aerophagia in this specific population and underreporting is possible. There are no data about the incidence of pathological aerophagia in the normal pediatric population. Most children without previous psychiatric diagnoses that present pathological aerophagia are dealing with acute stress situations (e.g. school entrance,

birth of siblings, divorce of parents, hospital admission). In those cases pathological aerophagia tends to disappear as the stressful situation resolves, but language and/or behavior problems are common, together with obvious signs of anxiety. In some patients pathological aerophagia is simply substituted by some other expression of anxiety.[6]

Some specialists even associate pathological aerophagia to self-destructing behavior. Mentally impaired children also suffer from anxiety, but its manifestations may be atypical and more difficult to detect and to treat in this population.

Pathological aerophagia may complicate with volvulus, intestinal necrosis/perforation and even death.[3,7,8] All reported surgical complications attained mentally impaired patients. Van der Kolk et al. describe nine severely retarded institutionalized adults with severe surgical complications (gastric perforation, gastric necrosis and volvulus, one death), which presented pathological aerophagia from childhood. Those authors suggest that untreated pathological aerophagia in neurologically impaired children may persist after maturity and lead to serious complications in the long-term, which may be prevented by the insertion of a gastrostomy.[9]

The disease might be caused either by psychogenic myoclonus-like air swallowing movements (as we observed in our index patient) or by repetitive reflex air swallowing movements due to paroxysmal openings of the upper esophageal sphincter in mentally impaired patients.

Pathological aerophagia patients normally present to the doctor because of severe persistent abdominal distension, typically minimal when the patient awakes in the morning (due to less air swallowing during sleep and elimination of flatus during the night period) and maximal at late evening. Associated abdominal pain is frequent. Belching/burping is also common. Audible air swallowing sounds are pathognomonic of the condition. Associated functional intestinal disorders have been reported, mainly constipation.[6] Occasionally there is some degree of respiratory restriction due to extreme abdominal distension. An abdominal radiograph shows gastric and diffuse intestinal distension, including small bowel, without signs of intestinal obstruction. Fluoroscopy may demonstrate the typical swallowing movements and abnormal pharyngoesophageal coordination.[10]

The differential diagnosis is with functional gastrointestinal problems (chronic intestinal pseudoobstruction, severe constipation, Hirschsprung's disease), malabsorption syndromes, anatomical intestinal obstruction and tracheoesophageal fistulae. Gas bloat syndrome may be considered in patients who had a previous fundoplication. The diagnosis of pathological aerophagia is rare and depends on clinical awareness. The exclusion of other conditions may demand complex complementary exams and delay treatment. In our case, the diagnosis was based on exclusion of intestinal muscular or neuronal abnormalities by biopsy, persistence of symptoms after control of constipation and presence of large quantities of air in the small intestine, without radiologic signs of intestinal obstruction (which exclude intestinal pseudoobstruction).

Little information concerning treatment of pathological aerophagia is available, with a very low level of evidence. Various pharmacological treatments have been suggested. Simethicone and anti-spasmodics can be tried, with low effectivity. Oral carbon tablets as air absorbents have been used, but their dosages and efficacy are unclear.[6,11] In the context of the good results of benzodiazepines to treat myoclonus-like syndromes and the observation that low dose intravenous clonazepan can abolish the pathological swallowing movements in pathological aerophagia patients, Hwang et al. prospectively treated 15 mentally impaired children with pathological aerophagia with a low oral dose of clonazepan (0.025mg/kg/day in two doses, slowly augmented to 0.1mg/kg/day as needed) for 4 months (or for a month after the patient was asymptomatic). The drug was slowly tapered off. No serious side effects were observed. When compared to a control group treated with only reassurance (n=7), 67% of the clonazepan group and 14% of the reassurance group resolved the symptoms (p=0.032), both without recurrence.[10] The effect of clonazepan may be centrally mediated, due to control of anxiety or affect directly the pathological mechanism that triggers the repetitive swallowing movements.

Behavioral/psychological therapy may be tried, but the efficacy is questionable in mentally impaired patients. Reassurance/psychotherapy was efficient in half of the cases treated by Delapetriere et al.[6] In our child, we tried specific pharmacological treatment for obsessive/compulsive disorder without success.

Phonoaudiologic treatment has been successfully tried in 3/13 patients[6] but is not an option in cases of serious psychiatric disease and/or cognitive deficit. In our patient speech therapists judged that reconditioning was not possible.

Decompression with a nasogastric tube can be used in cases of acute distension. Surgical treatments are rarely needed, almost exclusively in mentally impaired patients. Gastrostomy is the most frequent proposal, but may not always be effective, as in the cases of Fukuzawa and Gauderer.[3,12] Recently Fukuzawa et al.[3] proposed an esophagogastric separation and an abdominal esophageal decompressive stoma to treat two severely affected patients unresponsive to gastrostomy (both previously complicated with volvulus and necrosis).

In conclusion, pathological aerophagia is a rare condition that occurs in normal children exposed to high levels of stress, for whom the disease is normally self-limited, or in children with psychiatric or neurologic disease. In this last group, the disease may persist and cause serious complications. Pharmacological and behavioral treatments are ill-defined and their success is doubtful. Severe cases may demand surgical strategies, mainly decompressive gastrostomy.

Funding

This study did not receive funding.

Conflicts of interest

The authors declare no conflicts of interest.

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