CASE REPORT

Pathologic aerophagia: a rare cause of chronic abdominal distension

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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.

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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 deglutições 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 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.
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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); 102cm (<5th percentile of World Health Organization growth curve), 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 normo-fluid rectum. The rectum was full of solid feces.
Neuropsychiatric evaluation confirmed severe developmental delay and disproportionate aggressiveness toward the medical team. Several repetitive stereotyped move-
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Discussion

In children, 70% of the bowel gas comes from swallowed air. The radiological expression of aerophagia is the presence of gas throughout the small bowel, which is pathologic 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 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 ≥1 year at the time of diagnosis.

Psychiatric diseases attain 1/4 of pathological aerophagia cases, mostly mental retardation, autism and Rett syndrome. Although pathological aerophagia is rare, 8.8% of institutionalized mentally impaired children present aerophagia. 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.

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. 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.

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. 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 pharyngeoesophageal coordination.

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 effectiveness. Oral carbon tablets as air absorbents have been used, but their dosages and efficacy are unclear. In the context of the good results of benzodiazepines to treat myoclonus-like syndromes and the observation that low dose intravenous clonazepam 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 clonazepam (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 clonazepam group and 14% of the reassurance group resolved the symptoms (p=0.032), both without recurrence. The effect of clonazepam 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. In our child, we tried specific pharmacological treatment for obsessive/compulsive disorder without success.

Phonodiagnostic treatment has been successfully tried in 3/13 patients 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. Recently Fukuzawa et al. 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.

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Conflicts of interest
The authors declare no conflicts of interest.

References