Primary Achalasia: Practice Implications

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ABSTRACT

Achalasia is a rare but debilitating condition that affects approximately 1 to 2 in 100,000 persons. Although exact causes are unknown, achalasia may have its inception in familial, infectious, or autoimmune pathology. Regardless of the cause, patients with achalasia experience dysphagia, esophageal aparistalsis, regurgitation of food and fluids, and a range of other symptoms that impact quality of life for these patients. In this article, the authors explain the etiology and symptomatology of achalasia and present diagnostic and treatment options for practitioner consideration.

Keywords: achalasia, dysphagia, esophageal disease

Mrs. M. is a 47-year-old white woman with a medical history of hypercholesteremia and hypertension. She has been seen by her primary care physician every 6 months. During 1 of her routine visits, she reported experiencing intermittent episodes of feelings of food seeming to get caught in her throat while eating. The symptoms were empirically diagnosed as gastroesophageal reflux disease (GERD). Symptoms did not improve despite proton pump inhibitor and Reglan therapy (Robins Pharmaceuticals ltd, Auckland, New Zealand). Two years later, during an elective surgery for benign fibroid tumors, she aspirated and coded. A gastrointestinal consult postoperatively wrote that she had "Reglan failure" with severe GERD. Nine months later, Mrs. M. began experiencing frequent, spontaneous, and nocturnal regurgitation of undigested food. She had a greater than 50-lb unintentional weight loss. A barium swallow revealed a severely dilated esophagus with a *bird's beak* appearance to the lower esophageal sphincter (LES). Achalasia was confirmed by endoscopy later that month. Within days after the formal diagnosis was made, Mrs. M. was admitted to the hospital for severe substernal chest pain with radiation to the back, arms, and neck.

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The case study is typical of an achalasia presentation.[1] Achalasia is an esophageal motor neurodegenerative disease that frequently mimics other conditions, particularly in its early stages. The term *achalasia* is derived from the Greek, meaning "does not relax."[2] It is a rare and debilitating condition that affects approximately 1 to 2 in 100,000 persons.[3,4] Because of its potentially serious consequences, the nurse practitioner (NP) should consider it in the differential diagnosis of persons presenting with chest pain, dysphagia, or GERD unresponsive to treatment.[5] Prompt recognition of the problem will enable the NP to refer to the gastroenterologist in a more timely manner. This article examines the pathophysiology, etiology, symptomatology, differential diagnosis, and treatment of achalasia.

PATHOPHYSIOLOGY

Normal peristalsis results in the coordinated relaxation and contraction of the esophagus.[6] The muscles of the esophagus are innervated by the somatic efferent fibers of the vagus nerve at the proximal aspect and the preganglionic vagal nerve fibers along the distal aspect. Neurotransmitters such as nitrous oxide and vasoactive intestinal peptide along these pathways cause contraction and relaxation of the esophagus and LES. Persons with achalasia are noted to have an aperistalsis, which manifests as esophageal contractions that are either high amplitude and

simultaneous or low amplitude and nonprogressive.[7] This aperistalsis is caused by degenerative changes in the neurons along the esophageal wall.[6] The net result is an enlarged and often widely dilated esophagus with a narrowed LES.

ETIOLOGY OF ACHALASIA

Although exact causes are unknown, Park and Vaezi[6] postulated that achalasia may have its inception in familial, infectious, or autoimmune pathology. They noted that familial cases are extremely rare and are probably an autosomal recessive inheritance when they do occur. Infectious etiologies are more common. Chagas disease, caused by the protozoan parasite *Trypanosoma cruzi*, can cause incomplete LES relaxation and aperistalsis. There have also been case reports of achalasia-type symptoms associated with polio, Guillian-Barré, varicella, and viral esophageal infections. A large number of patients have an elevated antibody titer to varicella zoster and measles although this elevation is not present in all cases of achalasia.[8] Some autoimmune conditions such as systemic lupus, Sjogren syndrome, eosinophilic esophagitis, and scleroderma can develop HLA class II antigens that are often present in idiopathic achalasia. However, not all patients with achalasia have these HLA antigens, so causality cannot be established.[6] Boeckxstaens[9] proposed that antecedent trauma such as a motor vehicle accident or coronary artery bypass grafting may damage the vagal nerve, causing neuropathic dysfunction and leading to achalasia. Secondary achalasia can also be caused by diabetes mellitus or malignancy.[1,9]

Eosinophilic esophagitis may be a risk factor. It has been recently identified in approximately 1% of persons undergoing esophageal procedures verifying achalasia.[10] Eosinophilic esophagitis is an allergic reaction, resulting in eosinophils building up on the esophageal lining, causing inflammation and subsequent scarring, narrowing, and formation of excess fibrous tissue within the esophagus.[11] Mandaliya et al[10] studied the association of the 2 conditions, noting that when found together, poorer long-term outcomes were likely.

Allgrove (Triple A) syndrome is an extremely rare autosomal recessive disorder characterized by alacrima, achalasia, and adrenal insufficiency.[12] First described in the literature in 1978, onset varies between infancy and adulthood; the latter's symptoms generally manifest as progressive dysautonomia. Diagnosis of Allgrove syndrome is based on clinical examination and adrenal function testing. Further genetic testing confirms the diagnosis.[13] Because of its severe prognosis and high morbidity if left untreated, Allgrove syndrome should be considered in the differential for achalasia.

SYMPTOMATOLOGY OF ACHALASIA

Boeckxstaens[9] noted diagnostic delays, on average, of 5 years when establishing a diagnosis of achalasia. Most of the time, symptom presentation is typical but misinterpreted. The delay in diagnosis can also be related to early stages of the disease. Symptoms are frequently mild and/or can occur inconsistently. The most common symptoms are chest pain, GERD-like symptoms, and dysphagia. Chest pain generally occurs most frequently in younger patients and may present as substernal tightness or pressure with or without radiation to the arm or back. The chest pain frequently responds to sublingual nitroglycerin.[14]

Dysphagia is a hallmark of many esophageal conditions. There is a sensation of food or medications "sticking" in the throat or esophagus. A major symptom of achalasia will be a progressive dysphagia to both solids and liquids, with dysphagia to liquids being a key diagnostic finding. If dysphagia is the presenting symptom, it is wise to determine if the problem occurs during the actual act of swallowing or during the esophageal phase. The latter is more likely to be a clue to achalasia. Other symptoms may include regurgitation of food and fluids, nocturnal cough, reactive airway symptoms and choking while recumbent, heartburn, halitosis, frequent hiccups, and difficulty belching.[15,16] Some patients may be able to develop compensatory behaviors to aid in swallowing, such as throwing back the shoulders while eating to facilitate esophageal emptying.[14] Weight loss is generally mild, unless the disease is in its later stages. Screening for achalasia should occur for someone with recurrent episodes of aspiration pneumonia, bronchiectasis, or a finding of a lung abscess because of aspiration. Screening should also be undertaken for any immunocompetent individual presenting with oral candidiasis, which can occur