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Achalasia

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ABSTRACT

Achalasia is a rare neurogenic motility disorder of the esophagus, occurring in approximately 0.11 cases per 100,000 children. The combination of problems (aperistalsis, hypertensive lower esophageal sphincter (LES), and lack of receptive LES relaxation) results in patients having symptoms of progressive dysphagia, weight loss, and regurgitation. Treatment modalities have evolved over the past few decades from balloon dilation and botulinum toxin injection to laparoscopic Heller myotomy and endoscopic myotomy. Most data on achalasia management is extrapolated to children from adult experience. This article describes understanding of the pathogenesis and discusses newer therapeutic techniques as well as controversies in management.

Introduction and pathophysiology

Achalasia of the esophagus is a very rare condition, with an estimated annual incidence of 1:100,000 cases overall, and less than 5% of those occurring in children (0.11 per 100,000 pediatric patients).[1,2] It is found to occur more frequently in males. The condition was first described in 1672 by Sir Thomas Willis, and the term *achalasia* ("failure to relax") was coined by Hurt and Rake in 1929.[3] The neurogenic disorder is characterized by three abnormal findings in esophageal motility—absence of peristalsis, high resting pressure of the lower esophageal sphincter (LES), and failure of receptive relaxation of the LES. As a result, patients develop progressive dysphagia and regurgitation.

The etiology of achalasia is not fully understood, but it is considered to be due to an imbalance between the inhibitory and excitatory neurotransmitters in the distal esophagus secondary to loss of ganglion cells.[4] The absence of nonadrenergic, noncholinergic inhibitory ganglion cells leads to unopposed acetylcholine and substance P action resulting in a nonrelaxing LES.[3] Deficiency in nitric oxide releasing neurons in particular have been implicated in patients with achalasia.[3,5] Autopsy and resected specimens have also noted an inflammatory response with CD3/CD8-positive cytotoxic T cells as well as eosinophils and mast cells in the area, which may suggest an autoimmune phenomenon.[3,6,7] There are reports of hereditary and familial cases of achalasia pointing to a possible genetic link. Patients suffering from the "AAA" syndrome (*A*chalasia, *A*lacrima, *A*CTH insensitivity), or Algood's present with achalasia suggesting a potential genetic association in a small number of cases.[4] Associations with trisomy 21, congenital hypoventilation syndrome and glucocorticoid insufficiency have been reported.[8] Finally, there is an association with an infectious etiology with the parasite *Trypanosoma cruzi* causing aganglionosis of the LES in Chagas disease, prevalent in South America. The clinical findings in Chagas disease mirror that of typical idiopathic achalasia.

Achalasia is often discussed with other motility disorders of the esophagus such as diffuse esophageal spasm, or nutcracker esophagus, as they share some clinical and manometric features.[9,10] In some reports, it was suggested that diffuse spasm (DES) may progress to achalasia, however, this remains debated.[3,11] Some patients with spinal cord injury (SCI) are noted to develop esophageal motility disorders such as DES and achalasia, and these occur at a much higher rate than the non-SCI population.[11,12] Based on high-resolution manometric studies, there has been a division of achalasia into three subtypes based on the esophageal pressurization generated.[9] This has been found in adults, and may have implications on the therapy offered to some patients; however, it remains unclear and its existence in children is not known.[13]

Untreated cases of achalasia lead to an extremely dilated esophagus forming a tortuous, sigmoidal shape. This end-stage condition is known as mega esophagus and most treatment modalities are unsuccessful.[4]

Presentation and diagnosis

While adults and children both present with progressive dysphagia initially to solids and in some cases to liquids, the manifestations can be more protean and challenging to diagnose in children. A majority of pediatric patients, especially those

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younger than 6–7 years who present with difficulty in eating and progressive weight loss will be treated for failure to thrive and gastroesophageal reflux disease (GERD) due to the regurgitation, as those conditions are much more prevalent. Proton pump inhibitors, histamine receptor blockers, and prokinetics are used and the diagnosis may not be made for up to 6–10 years.[14] Some children are treated for feeding aversion due to the inability to eat foods with certain textures from progressive dysmotility and dysphagia.[8] Regurgitation of food in cases of achalasia is brought up without gastric contents or evidence of mixing, as opposed to severe GERD. Nocturnal cough is an important presenting feature in children and occurs due to chronic aspiration of esophageal contents when recumbent. Older children may present with chest pain from the dilation or acid exposure. A chest X-ray done for chronic cough may reveal the outline of a dilated esophagus and air–fluid level that can clue in to the diagnosis.

The diagnostic workup is similar to that for adults, with the exception that malignancy and pseudoachalasia is essentially nonexistent. Most clinicians start with an esophagram, which would show a dilated esophagus tapering into the LES in a "bird-beak" fashion (Figure). The degree of dilation is usually dependent on the duration of symptoms. A timed barium esophagram, which looks at the time to clear the esophagus, has been used as a definitive test by some authors as it can assess the peristaltic activity and the degree of LES hypertension as how long it takes to clear.[15,16] An upper endoscopy is a useful test to assess the mucosa and estimate the degree of LES hypertension. Biopsies can be performed to rule out other pathologies such as severe GERD. The gold standard test remains esophageal manometry, which involves placement of a catheter that measures the impedance during the act of swallowing and is able to assess all three critical components of the diagnosis of achalasia—peristalsis, resting LES pressure, and receptive LES relaxation. Some centers would still consider the diagnosis despite partial and a few normal LES relaxations due to some heterogeneity in children.[8,17] Manometry can also be performed to assess response to therapy as well as in recurrent symptoms. Additionally, intraoperative manometry has been utilized to help guide the length and completeness of the myotomy.[18,19] High-resolution manometry and intraluminal impedance monitoring are newer techniques that have been introduced. With high-resolution manometry, pressure plots are generated for the esophagus, which creates a topographical pressure map and helps to classify the condition into the three subtypes. This recent understanding was summarized in the Chicago classification of esophageal motility disorders, in which type I was defined as minimal esophageal contractility, type II as intermittent compartmentalized pressurizations with no peristalsis, and type III with spastic distal contractions.[10,20] High-resolution manometry may also help in situations where standardized manometry may not be completely diagnostic.[21]

The Eckardt score has been used in the past decade or more in adults, and it grades the symptom scores of dysphagia, regurgitation, chest pain, and weight loss on a 0–3 Likert-based scale.[22,23] Symptom scores can range from 0 to 12, and this score has been useful in monitoring success of therapy as well as in standardizing outcomes. Recently, this tool has been used in older children as well and has been useful.[16]

Medical management

Achalasia does not have a cure, and, therefore, all management principles are palliative to help the patient be able to swallow with less dysphagia. The purpose of any therapy for achalasia is improvement in esophageal emptying, usually achieved by reducing the LES pressures. There are no mechanisms to induce peristalsis in the esophagus; therefore, reduction in LES pressure is usually accompanied with dietary modification to allow gravity to assist esophageal clearance.

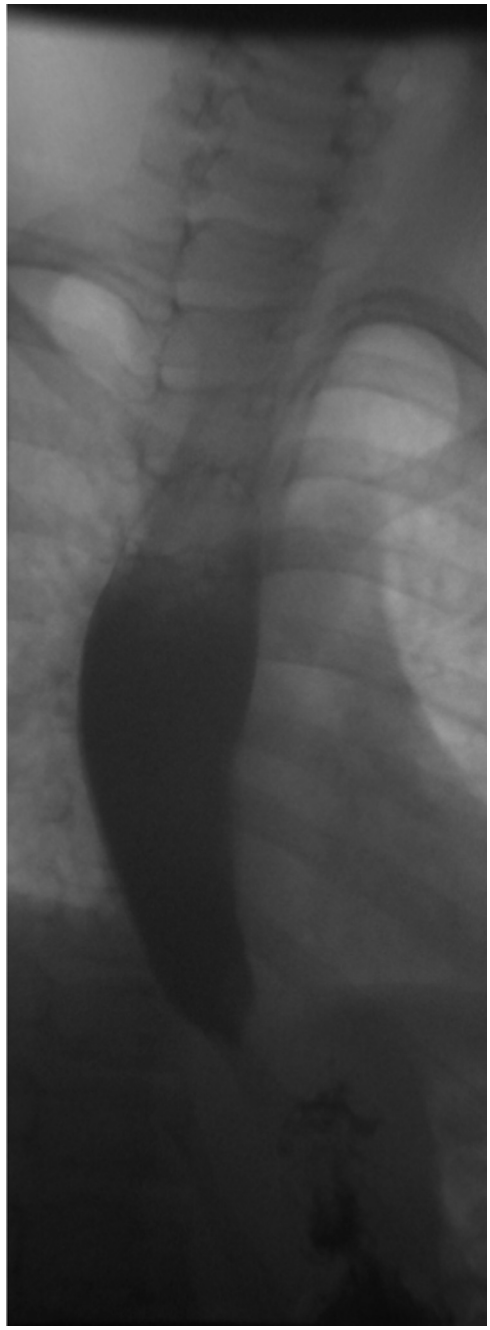

Fig. Esophagram with typical bird-beak appearance of distal esophagus.

Calcium channel blockers have been used in adults with some success but less is known about the effects in children. Nifedipine was used in a small report of 4 children who reported relief.[24] As reduced nitric oxide has been implicated in the pathogenesis of failure of relaxation, nitrates such as nitroglycerin have also been used, as has sildenafil, a nitric oxide potentiating agent.[25] The effects of all medications are very limited and should be used as a bridge to more definitive therapy, or in cases where any intervention would be considered too risky.

Dietary modifications are usually part of the natural adaptive process that occurs in each patient as the dysphagia progressively increases. These include a mostly liquid diet and frequent smaller meals with small bites. However, dietary modifications alone are