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ESOPHAGEAL ACHALASIA IN AN ADOLESCENT: A CASE REPORT

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Abstract

Achalasia is a rare disorder, particularly in Pediatrics, characterized by esophageal aperistalsis and inadequate relaxation of the lower esophageal sphincter. Its etiology remains unclear and is mostly idiopathic. This is a case of an adolescent male who presented with progressive dysphagia, occasional chest pain, and significant weight loss, and was diagnosed with Esophageal Achalasia through upper GI series, endoscopy, and manometry. The patient underwent Heller's Myotomy, the surgical gold standard for treatment.

Keywords: Achalasia, Heller's Myotomy, Esophageal Motility Disorder

Introduction

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 which occur in children (0.11 per 100,000 pediatric patients).[1] Pediatric achalasia is generally diagnosed between 7 and 15 years of age and has a slight predominance in boys.[2] In the Philippines, only 3 cases have been reported in the Philippine Pediatric Society Registry for the year 2020.[3] Achalasia is a pathological condition causing dysphagia, reflux, and regurgitation. The hallmarks of diagnosis include esophageal dysmotility and lack of relaxation of the lower esophageal sphincter (LES). Diagnosis is suspected by the clinical history, but is often delayed in children because of the rarity of the disease. If left untreated, the sequelae can be significant, as the proximal esophageal tissue becomes more compliant as a compensatory measure and entirely non-functional by end-stage disease. It is a life-long, debilitating condition, with a significant impact on quality of life. Hence, prompt diagnosis and intervention are needed. To date, pneumatic balloon dilation (PD) and Heller's myotomy (HM) are considered the most effective therapeutic options in children.[4]

Case Presentation

A 15-year-old male was admitted for a 1 year history of progressive dysphagia (initially to solids, and eventually progressing to liquids), with frequent regurgitation, occasional chest pain, and significant weight loss (~60 lbs or 27kg in 1 year). The patient had no known illnesses and this was the patient's first admission. The patient had normal vital signs for age and an unremarkable physical examination. Esophagoduodenoscopy was done for the patient revealing absence of any mechanical obstruction or inflammatory cause of the patient's esophageal symptoms. Gastric and distal esophageal biopsy showed unremarkable results with no active inflammation, intestinal metaplasia, atrophy, or dysplasia. Hence, an upper Gastrointestinal Series (UGIS) with barium contrast was facilitated. Results showed satisfactory swallowing mechanism, with no gross tracheal aspiration or nasopharyngeal regurgitation. However, there was narrowing at the region of the lower esophageal sphincter with lack of peristalsis, dilatation, and pooling of contrast at the distal segment of the esophagus. Findings were reflective of Achalasia (Figure 1). To further strengthen the diagnosis, an Esophageal Manometry test was done which showed incomplete relaxation of the LES in response to swallowing, high resting LES pressure, and absent esophageal peristalsis (Figure 2) which were consistent with a Classic Achalasia according to the Chicago Classification of Esophageal Motility Disorders for manometry (Figure 3). A chest CT scan was also done for the patient which showed dilated intrathoracic esophagus down to the gastro-esophageal junction with air-fluid level and non-thickened walls reflective of Achalasia (Figure 4). The patient then underwent Heller's Myotomy with Dor Fundoplication which the patient tolerated well with no immediate post-operative complications noted. The patient was admitted for a total of 7 days for post-operative care. The patient was initially placed on NPO for 24 hours post-operatively and feeding was slowly progressed until diet as tolerated was achieved prior to discharge. The patient had an unremarkable

hospital course with no noted regurgitation or chest pain post-operatively and was discharged well.

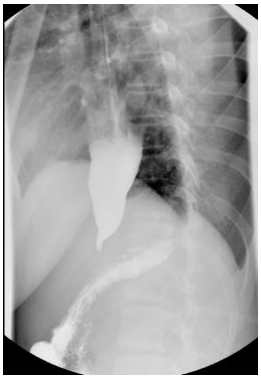

Figure 1. The patient's upper GI series result showing a narrowing at the region of the lower esophageal sphincter with dilatation of the distal segment of the esophagus— a classic "bird's beak" sign indicative of Achalasia.

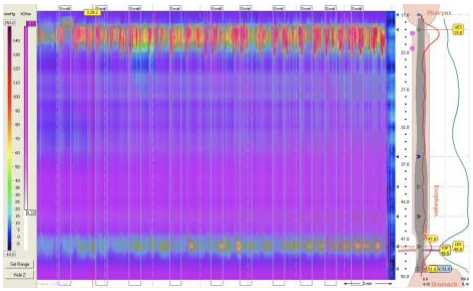

Figure 2. The patient's manometry result showing high resting LES pressure and absent esophageal peristalsis consistent with a Classic Achalasia according to the Chicago Classification of Esophageal Motility Disorders.

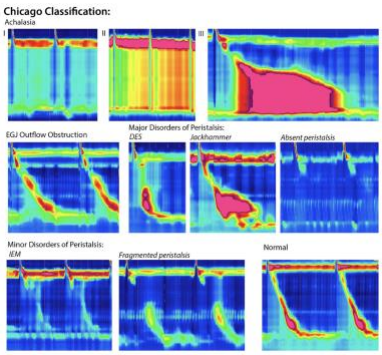

Figure 3. Examples of manometry color plots showing individual disorders based on the Chicago Classification for manometric findings.

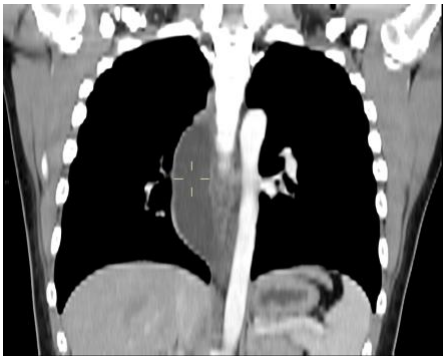

Figure 4. The patient's CT scan result showing a dilated intrathoracic esophagus down to the gastroesophageal junction

Differentials

The differential diagnosis of achalasia includes gastroesophageal reflux disease (GERD), especially in patients with chest pain of a burning quality typical of heartburn, other esophageal motility disorders, and pseudoachalasia due to a malignancy. These were all considered for our patient, hence the need for a complete workup. Here we note the similarities and differences of these conditions:

1. Gastroesophageal reflux disease – In patients with GERD, regurgitated food is typically sour tasting due to the presence of gastric acid. In contrast, in patients with achalasia, food and saliva are regurgitated from the esophagus and are therefore bland. Esophageal manometry is diagnostic of achalasia. Unlike the incomplete LES relaxation and aperistalsis that characterize achalasia, patients with GERD often have nonspecific manometric findings including ineffective esophageal motility and hypotensive LES.
2. Pseudoachalasia – Malignancy can cause pseudoachalasia either by invading the esophageal neural plexuses directly (eg, adenocarcinoma of the esophagogastric junction) or through the release of uncharacterized humoral factors that disrupt esophageal function as part of a paraneoplastic syndrome. In addition to gastric carcinoma, other tumors that can produce the syndrome include cancer of the esophagus, carcinoma of the lung, lymphoma, and pancreatic carcinoma. Patients with pseudoachalasia can have the same manometric findings as those with achalasia but can be differentiated by upper endoscopy and endoscopic ultrasound (EUS).

3. Other esophageal motility disorders – Patients with diffuse (distal) esophageal spasm and jackhammer esophagus may also present with dysphagia to solids and liquids. Esophageal manometry testing can distinguish achalasia from these esophageal motility disorders as LES relaxation (integrated relaxation pressure [IRP]) is normal in these conditions.

Discussion

Achalasia is a primary esophageal motility disorder characterized by the absence of esophageal peristalsis and impaired LES relaxation in response to swallowing. The LES is hypertensive in about 50% of patients. These abnormalities cause a functional obstruction at the gastroesophageal junction.[5]

The incidence of achalasia is approximately 1 per 100,000 people per year. In children, a retrospective study done from 1990 to 2013 from the Netherlands showed that the mean incidence of achalasia was 0.1 per 100,000 people per year.[6] In the Philippines, only 10 cases have been reported in the Philippine Pediatric Society Registry from 2019 to 2021 and only 3 were reported in the past year.[3]

The etiology of achalasia is largely unknown, however, there is some evidence that achalasia is an autoimmune disease. A European study compared immune-related deoxyribonucleic acid (DNA) in persons with achalasia with that of controls and found 33 single-nucleotide polymorphisms (SNPs) associated with achalasia. All of these were found in the major histocompatability complex region of chromosome 6, a location associated with autoimmune disorders such as multiple sclerosis, lupus, and type 1 diabetes.[7]

The pathophysiologic basis of achalasia is characterized by the degeneration of the inhibitory myenteric plexus that innervates the lower esophageal sphincter (LES) and esophageal body. This leads to an imbalance in the inhibitory and excitatory neurons resulting in the failure of the LES to relax with swallowing, absence of peristalsis of the esophageal body, and increased LES resting pressures.[8]

Achalasia is characterized by progressive dysphagia, regurgitation, chest pain, heart burn, and weight loss. In younger children and infants, they may also present atypically with recurrent pneumonia, nocturnal cough, aspiration, hoarseness, and feeding difficulties. Physical examination is usually normal for these patients.[8] Diagnosis is suspected by the clinical history, but is often delayed in children because of the rarity of the disease. Achalasia symptoms are often wrongly interpreted and contributed to GERD or an eating disorder, leading to delayed diagnosis and start of appropriate therapy. The Eckardt score was developed to objectify achalasia symptoms and is the sum of symptom scores for dysphagia, regurgitation, chest pain, and weight loss.[2] Each item is graded on a score of 0 to 3, with a maximum score of 12. Scores greater than or equal to 3 are considered suggestive of active achalasia (Table 1).[9] Symptoms alone, however, do not reliably diagnose the disease since there is an overlap of symptoms with other esophageal diseases, particularly gastroesophageal reflux disease. Furthermore, symptoms presence or severity does not correlate with manometric findings, degree of esophageal dilatation, or prognosis. A complete workup is necessary in these patients, not only for the diagnosis but for prognosis and to establish the proper therapeutic approach.[10] Our patient had a total Eckardt score of 8 (weight loss >10kg, daily dysphagia, occasional restrosternal pain, and daily regurgitation), which was indicative of active achalasia.

Table 1. Eckardt score for symptomatic evaluation in achalasia

Score	Weight loss (kg)	Dysphagia	Retrosternal Pain	Regurgitation
0	None	None	None	None
1	<5	Occasional	Occasional	Occasional
2	5-10	Daily	Daily	Daily
3	>10	Each meal	Each meal	Each meal

Esophageal manometry is required to establish the diagnosis. Diagnostic manometric findings of achalasia are incomplete relaxation of the lower esophageal sphincter and aperistalsis in the distal two-thirds of the esophagus. In patients with equivocal esophageal manometry results (eg, incomplete LES relaxation but some preserved peristalsis; some complete LES relaxation with aperistalsis), barium esophagram can help to assess esophageal emptying and esophagogastric junction morphology. Endoscopic evaluation with upper gastrointestinal endoscopy should be performed in patients with suspected achalasia to exclude a malignancy at the esophagogastric junction that can mimic achalasia.[4] Our patient underwent all these procedures, eventually leading to a diagnosis of achalasia. The patient's endoscopy was normal, ruling out pseudoachalasia from a malignancy. UGIS with barium contrast showed the classic "bird's beak" sign indicative of achalasia, and the diagnosis was confirmed by manometry showing high resting LES pressure and absent esophageal peristalsis consistent

with achalasia.

The diagnosis of achalasia always implies therapy. Goal of different therapies available for the treatment of achalasia is to reduce the LES pressure and improve bolus flow through the esophagogastric junction. Pharmacological treatment options, such as calcium channel blockers or endoscopic botulinum toxin injections in the LES are rarely used in the pediatric population because of their short-term effectiveness and potential for calcium channel blockers of side effects such as headache and dizziness.[11] To date, pneumatic balloon dilation and Heller's myotomy are considered the most effective therapeutic options in children. These therapies mechanically reduce the LES pressure by destruction of LES muscle fibers, either by stretching or cutting the LES. Postoperative GERD is a frequently reported complication after Heller's Myotomy, and therefore this procedure is most often combined with a partial fundoplication.[4] Our patient underwent Heller's Myotomy with Dor (or Partial Anterior) Fundoplication. Symptom improvement is reported in an average of 83% of patients undergoing transabdominal Heller Myotomy, pointing to an expected favorable outcome for the patient.[13]

Despite the effectiveness of currently available therapies for achalasia, 2–5% of patients are noted to develop what is called "end-stage" disease with a massively dilated esophagus and recurrent symptoms. In this group of patients, esophagectomy with reconstruction has been performed with reasonable outcomes. This is unlikely to occur in the pediatric age group, however, may happen with long-term follow-up of patients treated as children.[1]

While existing therapies aim at palliation through destruction of the LES, future treatment might aim to preserve this while addressing the main issue of absence of the myenteric ganglion cells. There has been one case report of using steroids in achalasia to arrest the loss of ganglion cells. However, in most patients the cells are already gone at the time of diagnosis.[12] Stem cell-based treatments for achalasia may also hold promise in the future as research has shown the development of gut-differentiated neural cells.[14]

Conclusion

Achalasia is a rare condition affecting esophageal motility in children. In a manner similar to the disease found in the adult population, children experience symptoms of dysphagia, regurgitation, and chest pain due to a failure of relaxation of the lower esophageal sphincter. Standard diagnostic approaches include upper endoscopy and esophageal manometry. Achalasia therapy is aimed at improving esophageal emptying by reducing lower esophageal sphincter tone either pharmacologically or surgically. However, there is currently no defined therapeutic algorithm for the pediatric population.[15] Guidelines for pediatric achalasia rely upon a combination of data obtained from studies in adults and expert opinion. Hence, there is a need for well-designed intervention trials in children with achalasia to determine optimal management and facilitate evidence-based clinical guideline development.

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