Case Report

Achalasia – A Case Report of Rare Dysphagia in a Paediatric Patient

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Abstract
Achalasia is a rare neurodegenerative disorder of the oesophagus characterised by failure of lower oesophageal sphincter (LES) relaxation. Achalasia is a rare disease in children which presents with symptoms that can mimic a variety of common paediatric illnesses.

A 15 years old girl was admitted to the paediatric clinic with complaints of difficulty swallowing solid foods and liquids, chest tightness after eating, followed by regurgitation of undigested food late at night. The patient had the symptoms for the past 2 years with the last 2 months becoming progressively worse. A barium swallow, an upper endoscopy and an endoscopic ultrasound were performed which confirmed the diagnosis. After a consultation with a paediatric surgeon, the patient was referred to the Surgery clinic for a planned surgical intervention.

The authors present a review of the literature in relation to an achalasia case observed at their clinic.

Keywords: achalasia, dysphagia, children

Introduction
Achalasia is a rare dysmotility disorder of the oesophagus with an incidence of approximately 1:100,000 patients, with an equal gender distribution. Less than 5% of symptomatic patients present under the age of 15. Although the disease is most frequently idiopathic in nature, it has been associated with Trisomy 21, eosinophilic esophagitis, Chagas’ disease, Allgrove syndrome etc. [1].

Children most often present with symptoms of progressive dysphagia, vomiting and weight loss. Younger patients may present atypically with refractory cough, hoarseness, aspiration, recurrent pneumonia, sinusitis, bronchial asthma, eating difficulties and weight loss. Because these conditions are more frequently seen in gastroesophageal reflux disease (GERD), the diagnosis of achalasia is often delayed by a few months up to 10 years after the initial presenting symptoms [2].

Case Presentation
History of present Illness
Patient presentation: A 15 years old female was referred to the paediatric clinic after consultation with a paediatric gastroenterologist. Her presenting complaints were heart burn and abdominal discomfort which started 2 years ago. On closer examination she revealed to have dysphagia for solid foods and liquids, “feeling of congestion” during the act of swallowing, severe chest pain and regurgitation of partially digested foul, smelling food late at night.
with the symptoms becoming more severe in the past 2 months. She had had an X-ray oesophagogram which was consistent with GERD.

**Past medical history**

This was the mother’s second pregnancy with the first pregnancy ending in an elective induced abortion. The prenatal history of the mother was unremarkable excluding an infection of the genitourinary tract at around 28 weeks of gestation. There were no known complications at birth, the delivery was a normal vaginal delivery, and birth occurred at full term. The patient’s birth weight was 3450g and height 48cm. The patient’s past medical history includes a tonsillectomy at 5 years of age and an infection of the genitourinary tract during early childhood. The patient had all the required childhood immunizations. Family history: The patient’s mother was suffering from gastritis and allergies; Her father had been diagnosed with peptic ulcer disease and hypertension.

**Medications**

The patient had been taking a protein pump inhibitor, domperidone, and a probiotic prior her hospitalization.

**Physical examination**

The patient was a young relatively healthy appearing girl. Height was 162cm and weight 54kg, BMI 20, afebrile. Skin was pink with no visible rash, normal turgor and elasticity, body fat normally expressed. Lymph nodes were not palpable. Her cardiovascular and pulmonary examinations were normal. Her abdomen was flat with active bowel sounds. It was soft and tender to deep palpation of the epigastrium; there were no palpable masses or hepatomegaly. Her neurological and musculoskeletal examinations were also normal.

**Management**

The patient was treated with H$_2$ - antagonists, a probiotic and venous infusions at admittance. Her laboratory results were normal. During the patient’s stay she underwent a series of diagnostic tests. A barium swallow was performed which showed a severely dilated oesophagus (megaesophagus) with a stricture at the lower oesophageal sphincter with low-amplitude peristalsis (Figures 1 & 2).

![Figure 1: Contrast radiography showing stricture at the LES](image-url)
There were no signs of defects or irregularities of the mucosal folds. An upper endoscopy was also performed which showed a dilated oesophagus with retention of partially digested food and retained liquid without visible macroscopic damage of the mucosa (Figure 3).

An endoscopic ultrasound was performed which showed thickening of the lower oesophageal sphincter which extends 5 cm upward followed by marked thinning of the oesophagus. The diagnostic results and clinical picture were supportive of the diagnosis achalasia. After a consultation with a paediatric surgeon, the patient was referred to the Surgery clinic for a planned surgical intervention.

**Discussion**

**Epidemiology and pathology**

Achalasia is a rare disease in the paediatric population. As previously cited it is estimated to affect 0.18/100,000 children annually [1]. The disease may present in infants as young as 7 weeks of age, but overall it
extremely rarely presents in children younger than 15 years old. As in adults, paediatric achalasia is most commonly idiopathic and sporadic in nature; nevertheless, the disease may be associated with genetic and familial conditions.

Familial achalasia is extremely rare, having been reported in parent and child with achalasia and multiple siblings from consanguineous relationships suggesting an autosomal recessive inheritance. Achalasia may also be associated with multiple syndromes such as triple-A syndrome (achalasia, alacrimia, addisonism) also called Allgrove’s syndrome, Sjogren’s syndrome and Down’s syndrome [1].

The characteristic pathologic hallmark of idiopathic achalasia is inflammation and destruction of inhibitory myenteric ganglion cells of the oesophagus. This was documented by Goldblum et al. who studied the oesophagi of 42 achalasia patients after undergoing total thoracic oesophagectomy. They found reduced myenteric ganglion cells within the oesophageal body in all of the specimens and total absence in 20 of them [3]. The exact initial offending agents leading to inflammation of the myenteric plexus remain unknown with viral infections being the most cited.

The resulting oesophageal aperistalsis and incomplete relaxation of the LES lead to impaction and accumulation of food in the oesophagus. Over time this results in progressive dilation of the body of the oesophagus.

Clinical presentation

The symptoms of achalasia in adolescents and adults are nonspecific: dysphagia (95%), regurgitation of undigested food and aspiration (60-70%), heartburn (40%) and chest pain [1]. These symptoms progress over time which makes for a challenging diagnosis. The Eckardt score (Table 1) is the most commonly used scoring system which aids in reaching the diagnosis in adolescents and adults. It is the sum of the scores for dysphagia, regurgitation, and chest pain. The maximum score on the Eckardt scale is 12.

Table 1: Eckardt scale and scoring system

<table>
<thead>
<tr>
<th>Symptom</th>
<th>Each meal</th>
<th>Daily</th>
<th>Weekly</th>
<th>None</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dysphagia</td>
<td>3</td>
<td>2</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td>Regurgitation</td>
<td>3</td>
<td>2</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td>Chest pain</td>
<td>3</td>
<td>2</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td>Weight loss</td>
<td>&gt;10kg</td>
<td>5-10kg</td>
<td>&lt;5kg</td>
<td>No weight loss</td>
</tr>
</tbody>
</table>

In younger patient’s achalasia may present with atypical symptoms. The rarity of achalasia in children and the presence of symptoms that mimic other common paediatric illnesses may lead to misdiagnosis in these patients [1]. The most common misdiagnosis is GERD which can also present with failure to thrive, feeding difficulties, and recurrent respiratory tract infections. Given the frequency of GERD in the paediatric population in relation to achalasia, around 50% of children are treated with prokinetic and antacid medications until a definitive diagnosis of achalasia is reached. This had been outlined by Hallal et al. who studied 13 patients diagnosed with achalasia, 6 of whom had been treated for GERD and asthma [2]. Common symptoms have been implicated in reports of patients diagnosed with eating disorders such as anorexia nervosa and bulimia that were subsequently diagnosed with achalasia and successfully treated [1]. All of this highlights the importance of clinical alertness in cases of dysphagia associated with other disorders.

Diagnostic workup

Achalasia is diagnosed with a barium swallow study which is then confirmed with oesophageal manometry, upper endoscopy and other modalities. Barium swallow studies typically demonstrate a dilated esophagus proximally...
to a “bird’s beak” like tapering of the distal oesophagus. The esophagogram study is often diagnostic because of the delay in diagnosis in many paediatric patients.

The gold standard in achalasia diagnosis is oesophageal manometry which typically demonstrates increased LES resting pressures, abnormal oesophageal peristalsis and relaxation of the LES during the act of swallowing [4]. The diagnosis cannot be excluded in the absence of the aforementioned findings because LES function in children is heterogeneous. According to Morera et al. partial relaxations are common and normal relaxation may also be present on manometry in paediatric achalasia patients [5].

Upper endoscopy with oesophageal biopsy is a useful modality which should be used to rule out other possible causes of pseudoachalasia such as eosinophilic oesophagitis, malignancy, mechanical structures or rings, candidal infections and advanced GERD [1].

Other diagnostic modalities can be used in cases where the diagnosis is not certain such as MRI scan, oesophageal radionucleotide studies, and endoscopic ultrasound. All of them can be useful in the diagnostic process as well as in monitoring the subsequent treatment.

**Treatment:** There are currently no definitive guidelines for treating achalasia in the paediatric population. The main objective in the treatment of achalasia is to reduce LES pressure in order to facilitate oesophageal emptying by either medical therapy or surgical techniques.

**Medical therapy:** Medical therapy, while not used as a definitive treatment, may be used as a bridge to surgical therapy. Even though calcium channel blockers such as nifedipine have been used in adults, there is limited medical information to support their use in children. Nifedipine have been used in the treatment of four adolescent patients who showed significant improvement in symptoms, while other sources report that the side effects of the medication outweigh the benefits in this age group and thus cannot be recommended as first line treatment for symptom relief.

There are several endoscopic techniques that are used in the treatment of Achalasia. Botulinum toxin inhibits the release of acetylcholine by the presynaptic nerve cells resulting in muscle relaxation. These properties have been used in relieving the symptoms of achalasia by injecting the toxin into the LES. The procedure is fairly simple to perform, with very few complications. Unfortunately, very few paediatric patients remain symptomless with many of them still requiring endoscopic ballon dilatation or myotomy of the LES after 4-6 months [6].

Endoscopic balloon dilation is a well-tolerated procedure with relatively few risks in adult patients with achalasia. Similar results have been documented in children with 65-80% of patients treated with balloon dilation reporting no symptoms in 2 to 8 years after the procedure [6]. The most often complications of the procedure include GERD, retrosternal or epigastric pain, and rupture of the oesophagus. The risk of oesophageal rupture after the procedure is reported to be 6%, according to a study that documented 50 children who underwent balloon dilation of the LES. Even though the procedure has been performed successfully on patients as young as 7 weeks old, balloon dilation is not recommended in children younger than 9 years of age due to technical limitations and increased risk of complications [1]. Despite recurrence of symptoms, balloon dilation has proven to be a cost effective, relatively low risk procedure for achalasia treatment. Nevertheless, patients in need of more than 1 dilation over the course of a year should be considered for surgical myotomy.

Although currently there are no algorithms for the treatment of achalasia in children, comparison between reports of balloon dilation and Heller myotomy has revealed that Heller myotomy is the procedure of choice based on symptoms recurrence and need for subsequent intervention [6].
Surgical procedures: Heller first introduced the cardiomycotomy in achalasia patients which includes separation of the LES from the oesophageal wall with the extension interiorly over the first 2 cm of the gastric cardia. Originally the procedure was done through a laparotomy which has largely been replaced by the minimally invasive Laparoscopic Heller Myotomy (LHM) with or without an antireflux procedure [7]. The benefits of LHM include shorter hospital stay, decreased postoperative pain and faster return to daily activity. The procedure is not without its risks potentially including intraoperative or postoperative oesophageal perforation, recurrent dysphagia, GERD and insufficiency of the myotomy resulting in the need for a repeat of the surgical intervention or balloon dilation [8]. Multiple centers recommend the use of intra-operative manometry or endoscopy to limit the chance of the aforementioned complications.

Currently the combination of LHM with an antireflux procedure is debatable. Although several single case studies did not show higher incidence of reflux in paediatric patients treated with LHM alone, the majority of studies suggest that LHM with fundoplication is superior to LHM alone in preventing postoperative GERD. At this time surgery is considered to be the only definitive treatment of achalasia.

PerOral Endoscopic Myotomy (POEM) is a relatively new technique which is gathering popularity as an alternative to LHM or balloon dilation in adults as well as in children. The procedure is done by creating a submucosal tunnel into the oesophageal wall by carbon dioxide (CO₂) insufflations and coagulation, after which a longitudinal myotomy of the circular oesophageal musculature is performed. Reported complications include subcutaneous emphysema, mucosal injury, pneumothorax, pneumomediastinum, pleural effusion, and pneumoperitoneum [1]. The incidence of these gas related complications has been significantly reduced by replacing air with CO₂ because of its better diffusion and easier complication management with conservative measures. The main disadvantage of POEM in the treatment of achalasia is the higher incidence of postoperative GERD compared to LHM with fundoplication. Studies suggest that a third of adult patients who underwent POEM developed GERD according to 24h pH studies [1].

Recent retrospective studies examined the results of paediatric patients who underwent LHM with Dor fundoplication compared with the POEM procedure, which showed comparable improvement in symptoms in both groups [1]. In addition, POEM patients experienced less pain, took fewer medications, and showed earlier return to daily activity. Unfortunately, much of the data collected on the outcomes of POEM is retrospective with short follow-up period and yet unknown long-term efficacy.

Conclusion

Achalasia is a rare disease in the paediatric population and every documented case further advances the diagnostic and therapeutic outcomes in these patients. Even though the diagnostic methods have been established, the therapeutic approach at this time depends on each individual patient and the experience of his or her clinician.

Nevertheless, more data is needed to formulate definitive guidelines for the diagnosis and treatment of paediatric achalasia.

References


