Review

Achalasia in a child: A case report and review of literature

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Achalasia is a rare neurodegenerative disease of the oesophagus in paediatric population. This age group can present with typical symptoms of the disease like dysphagia, chest pain, vomiting, belching, regurgitation of undigested food and failure to thrive but occasionally can present with a typical symptoms like recurrent pneumonia, nocturnal cough, aspiration, hoarseness, and feeding difficulties thereby making diagnosis very difficult. We report a case of achalasia in a child that presented with a typical symptom. Our patient received various anti-tuberculous drugs and anti-asthmatics with no significant improvement. Barium swallow revealed a dilated distal oesophagus with a smooth tapering. A diagnosis of achalasia was made and he had modified Hellers procedure. We reviewed relevant literature to update physicians on this disease entity.

Key words: Achalasia, oesophagus, childhood, neurodegenerative disease.

INTRODUCTION

Achalasia is caused by loss of inhibitory innervation of lower esophageal sphincter and is characterized by failure of the sphincter to relax. This failure of relaxation causes poor emptying of the esophagus and subsequent dilatation and abnormal contractility of the proximal esophagus (Mehdi et al., 2008). It is a rare esophageal neurodegenerative disorder in the pediatric population. The disease is even more infrequent in children less than 5 years of age. The incidence of achalasia in childhood is 0.11/100000 children annually. Overall, less than 5% of patients are with symptoms present under the age of 15 (Franklin et al., 2014; Hamza et al., 1999). The most commonly presenting symptoms of achalasia include dysphagia, chest pain, vomiting, belching, regurgitation of undigested food and failure to thrive. Younger children and infants may also present a typically with recurrent pneumonia, nocturnal cough, aspiration, hoarseness, and feeding difficulties as such making the diagnosis extremely difficult (Morera and Nurko, 2012). Due to its rare occurrence, achalasia is not commonly thought of in evaluating children with above symptoms, and diagnosis can be consequently delayed.

CASE REPORT

A 12 year old male child that presented with dysphagia and regurgitation of 4 years duration. There was cough, chest pain, belching and weight loss but no history of contact with a chronically coughing patient. He was said to have received various medication for asthma and tuberculosis with no significant improvement and no history suggestive of ingestion of foreign body or corrosive agent. He was found to be wasted otherwise other findings were normal.

Barium swallow revealed a dilated oesophagus with smooth tapering of the distal end (Figure 1). Endoscopy showed dilated oesophagus with retained food debris. A diagnosis of achalasia cardia was made and was treated for respiratory tract infection and subsequently worked up for surgery. He had modified Hellers procedure via an abdominal route. Postoperative recovery was uneventful.
and was discharged on the 7th day post op.

REVIEW OF LITERATURE

Historical perspective

In 1674, Thomas Willis first described this disease as "The mouth of the stomach being always closed, either by a tumor or palsy, nothing could be admitted into the ventricle unless it was violently opened" (Zhang et al., 2009).

In 1881, von Mikulicz described the disease as a cardio spasm to indicate that the symptoms were due to a functional problem rather than a mechanical one. In 1929, Hurt and Rake realized that the disease was caused by a failure of the lower esophageal sphincter (LES) to relax. They coined the term achalasia, meaning failure to relax. Thomas in 1969 described cricopharyngeus achalasia in infants, which may develop between birth and 6 months (Utian and Thomas, 1969; Jain and Bhatnagar, 2009; Sari et al., 2007).

Epidemiology

The disease is more prevalent in males and is most commonly idiopathic. Incidence is 0.11/100000 children annually.

Aetiology

There has been much debate over the aetiology of achalasia with several potential triggers for the inflammatory destruction of inhibitory neurons in the oesophageal myenteric plexus being implicated. These include autoimmune responses, infectious agents and genetic factors (O’Neill et al., 2013; Hallal et al., 2012).

Autoimmune conditions

Patients with achalasia are 3.6 times more likely to suffer an autoimmune condition, compared with the general population. Sjogren’s syndrome, Systemic Lupus Erythematosus and uveitis were all significantly more prevalent in achalasia patients. The study also found the presence of a T-cell infiltrate and antibodies within the myenteric plexus of many patients with achalasia and an increased presence of human leukocyte antigen class II antigens (O’Neill et al., 2013; Gockel et al., 2014).
**Infectious agents**

Several infectious agents have been implicated. Chagas disease has a known infectious aetiology, and exhibits many similarities with achalasia (Ghoshal et al., 2012). In addition, there are several reports of varicella zoster virus and Guillain-Barre syndrome preceding the onset of achalasia. Antibody studies have demonstrated increased titres to herpes and measles viruses in patients with achalasia in comparison to healthy control groups. One study looking specifically at the link between the herpes simplex virus (HSV) and primary achalasia indicated the presence of HSV-1 reactive immune cells in the lower oesophageal sphincter of achalasia patients, suggesting that HSV-1 may be involved in the neuronal damage to the myenteric plexus leading to achalasia (Lau et al., 2010; Birgisson et al., 1997).

**Autoimmune predisposition**

The genetic basis for achalasia has not been widely investigated due to its low prevalence. One syndrome, known as the triple “A” syndrome, which consists of a triad of achalasia, alacrima and adrenocorticotropic hormone resistant adrenal insufficiency, is a known autosomal recessive disorder caused by gene mutations on chromosome 12. This syndrome, together with the prevalence of cases within children of consanguineous couples suggests the possibility for a genetic component to the aetiology of achalasia. Associations have been reported with other genetic diseases including Parkinson’s disease, Down’s syndrome and MEN2B syndrome (Nihoul-Fekete et al., 1991; Myers et al., 1994). One recent study suggested the possibility of involvement of the rearranged during transfection gene, which is a major susceptibility gene for Hirschsprung’s disease (also linked with Down’s syndrome; Park and Vaezi, 2005).

It has been postulated that achalasia may incorporate a multi-factorial aetiology with an initiating event such as a viral or environmental insult resulting in oesophageal myenteric plexus inflammation. This inflammatory reaction may then initiate an autoimmune response in a susceptible group of genetically predisposed people, causing destruction of inhibitory neurons (O’Neill et al., 2013; Chuah et al., 2012; Booy et al., 2012; Kaar et al., 1991; Park and Vaezi, 2005).

**Pathophysiology**

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 Pressure (Franlin et al., 2014; Gockel et al., 2012). Lack of peristalsis and a non-relaxing lower oesophageal sphincter cause progressive dysphagia. Regurgitation, particularly at night, with aspiration of undigested food and weight loss can be presenting features, particularly in established disease. Features, which present in the early stages of the disease, may be similar to that of gastro-oesophageal reflux, including retrosternal chest pain typically after eating and heartburn (Francis and Katzka, 2012). Due to low prevalence of achalasia especially in children, the condition is undiagnosed and usually present lately. In children, regurgitation and aspiration results in features of respiratory tract infection. These symptoms are also mistaken for reflux oesophagitis.

**Clinical features**

The most frequent symptoms of achalasia are dysphagia, chest pain, regurgitation of food, and weight loss. Secondary pulmonary disease can occur due to regurgitation and aspiration of retained esophageal contents. This can cause symptoms of chronic cough, especially nocturnal cough, choking, recurrent pulmonary infections, pneumonia, wheezing, atelectasis and rarely pulmonary empyema. Some patients may develop hoarseness of the voice caused by direct pressure of distended esophagus on the recurrent laryngeal nerve. Tracheal obstruction due to compression from dilated esophagus may occur in achalasia and can be the only presentation. This can be a serious and potentially life threatening complication of achalasia (Mehdi et al., 2008; Kugelman et al., 2000; Akhter et al., 1988; Chapman et al., 1989; Lee et al., 2010).

**Diagnosis**

**Chest Radiograph**

Preliminary chest radiograph in patients with achalasia will show dilated oesophagus, which will manifest as widened mediastinum if its long-standing, air-fluid level is seen on a lateral film, there may be pulmonary infiltrates depicting aspiration pneumonitis and there may be absent gastric air bubble.

**Barium studies**

Barium swallow studies classically will demonstrate a dilated esophagus with “bird’s-beak” like tapering of the distal esophagus. Often, since there is a significant delay in diagnosis of achalasia in children, the esophagram study alone is diagnostic (Walzer and Hirano, 2008).


**Oesophageal manometry**

This may reveal elevated resting lower oesophageal sphincter (LES) pressure, absent or low amplitude peristalsis, or non-relaxing LES upon swallowing are diagnostic findings in children with achalasia (Lelli et al., 1997).

**Endoscopy**

Upper gastrointestinal endoscopy and biopsy is reasonable to rule out esophagitis, Trypanosoma cruzi, malignancy, and other secondary causes of Achalasia (Franlin et al., 2014; Hamza et al., 1999).

**Treatment**

The various methods of treatment of achalasia involve reduction of LES pressure in order to facilitate esophageal emptying by: injection of botulinum toxin, oral administration of calcium channel blockers (Nifedipine), pneumatic dilatation, or esophageal myotomy (Heller) with or without an anti-reflux procedure (Franklin et al., 2014; Patti et al., 2001; Maksimak et al., 1986).

**Medical treatment**

The use of calcium blocking agents has not been extensively studied in children. A report of 4 children treated with nifedipine reported relief of symptoms likely related to a decrease in resting LES pressure (Babu et al., 2001).

Endoscopic injection of Botulinium toxin into the LES has its optimal dosing and injection frequency of botulinum toxin to relieve achalasia symptoms in children has not been well defined as such its use in children is limited (Franklin et al., 2014).

**Pneumatic dilatation**

This has been reported in children. Multiple dilatations are often required to achieve successful relief of symptoms although initial response predicts the success or failure of subsequent dilatations. The advantages of balloon dilatation include shorter length of stay, quicker recovery time, and decreased cost. Multiples studies reported successful outcome especially in older children (Boyle et al., 1981; Nakayama et al., 1987; Mehta et al., 2005; Ortiz et al., 2008).

**Surgical treatment**

Surgery has been reported to be the most definitive and successful treatment of choice (Franklin et al., 2014; Salvador et al., 2014). Surgery can be open or laparoscopic/thoracoscopic. The approach can be either abdominal or thoracic. The transthoracic approach with video allows the myotomy to be made with sufficient length, but does not allow the anti-reflux procedure to be performed. Postoperative pH testing reveals that transthoracic myotomies present a 60% level of gastroesophageal reflux complication. The abdominal laparoscopic approach allows the myectomy to be performed in association with the anti-reflux Procedure (Patti et al., 2001; Corda et al., 2010; Rothenberg et al., 2001; Rosemurgy et al., 2010).

Laparoscopic Heller’s myotomy is the treatment of choice in both adults and paediatrics. It involves a longitudinal incision in the muscle of the esophagus approximately 5 cm above the esophagogastric junction and extending 2-3 cm onto the cardia of the stomach (Walzer and Hirano, 2008; Patti et al., 2001; Liu et al., 2004).

Per oral endoscopic myotomy (POEM)- is one of few procedures utilizing natural orifice transluminal endoscopic surgery (NOTES) routinely in adults. POEM is an endoscopic procedure that directly treats the diseased tissue (Franklin et al., 2014; Rothenberg et al., 2001). It is performed utilizing flexible endoscopy, mucosal incision and dissection of a submucosal tunnel distally in the esophageal wall to approach the esophagogastric junction. A 2-3 cm longitudinal incision in the inner circular muscle approximately 4 cm from the LES, will produce similar results to Heller myotomy (Franklin et al., 2014; Friedel et al., 2013). A contrast esophagram is routinely obtained on the first postoperative day and the patient is started on a pureed diet if esophagram is normal (Franklin et al., 2014).

**CONCLUSION**

Achalasia is not common in children as such can pose a diagnostic challenge. A high index of suspicion is required to make the diagnosis and appropriately treat this group of patients. Outcome of surgical treatment is good and POEM has been shown to be effective in children.

**REFERENCES**


