


Achalasia in a Female Adolescent

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ABSTRACT

Achalasia is an esophageal smooth muscle motility disorder due to relaxation failure of the lower esophageal sphincter. This condition is rarely found in children; 2-5% cases occur in less than 16-year old. **Case**: A 16-year-old girl presented with chief complaint of regurgitation. She also complained of dysphagia, weight loss, and general weakness. Her general appearance was weak, underweight, and decreased skin turgor. Barium esophagogram demonstrated a narrowing of esophagus lumen distal to the esophagogastric junction with a "bird's beak" appearance or rat tail sign. Esophageal achalasia was diagnosed and managed with open (laparotomy) surgical myotomy and fundoplication. Post-operative diet arrangement was carried out gradually and the patient was discharged 4 days post-surgery.

Keywords: Achalasia, esophageal smooth muscle, motility disorder

ABSTRAK

Akalasia merupakan gangguan motilitas otot polos esofagus yang terjadi akibat kegagalan relaksasi *lower esophageal sphincter* (LES). Kondisi ini jarang ditemukan pada anak-anak; 2-5% kasus pada anak berusia di bawah 16 tahun. **Kasus**: Seorang remaja perempuan 16 tahun datang ke poliklinik bedah anak dengan keluhan utama regurgitasi. Anak juga mengeluh disfagia, penurunan berat badan, dan kelemahan umum. Keadaan umum anak lemah, berat badan rendah, dan penurunan turgor kulit. Esofagogram barium menunjukkan penyempitan lumen esofagus distal terhadap *esophagogastric junction* dengan tampakan "*bird's beak*" atau tanda *rat tail*. Akalasia esofagus didiagnosis dan ditatalaksana dengan pembedahan laparotomi miotomi dan fundoplikasi. Pemulihan diet pasca-operasi secara perlahan dan pasien dipulangkan 4 hari pasca pembedahan. **Kukuh Rizwido Prasetyo, Yudi Suryana. Akalasia pada Seorang Remaja Perempuan.**

Kata kunci: Achalasia, otot polos esofagus, gangguan motilitas

INTRODUCTION

Achalasia is an esophageal smooth muscle motility disorder due to failure of relaxation of the lower esophageal sphincter, causing a functional obstruction at the gastroesophageal junction. This condition was first described by Sir Thomas Willis in 1672.[1] Achalasia is very rare, with an annual incidence of roughly 1 per 100,000 people and a prevalence of 10 per 100,000.[2] Achalasia occurs with equal frequency in both males and females; mainly found in adult between second to fifth decade of life, with a peak incidence between 30 to 60 year-old.[3] It is very rare among children; less than 2% to 5% cases occur in children less than age 16.[1]

The exact etiology of neural degeneration in achalasia is unclear though many theories have been proposed, including autoimmune mechanism, viral infection, and genetic predisposition.[1] Patients present with a wide range of symptoms, depending on age and stage of the disease. Since the cause is still not fully understood, treatment remains symptomatic, with pharmacological, endoscopic and surgical choices.[4]

A case of 16-year-old female with achalasia was presented, with discussion on its epidemiology, etiopathogenesis, clinical presentation, investigations, and treatment in pediatric population.

CASE

A 16-year-old girl was brought by her family to our Pediatric Surgery Clinic with chief complaint of persistent vomiting, resulting in general weakness. The patient complained of swallowing difficulty (dysphagia) since the age of 10 years, The symptom worsened with more difficulty in swallowing, nausea, epigastric pain and burning, and weight loss. The patient had seen doctor several times and was treated for stomach problems, but the symptoms got worse until a few months before admission, she could not swallow food and drink entirely; all food and drinks were vomited in original shape, consistency, and color. The patient lost weight and felt weaker, also felt unwell and 'feverish'. She denied any shortness of breath or choking.

Her general appearance was thin and weak but still alert. Vital signs were: blood pressure 100/70 mmHg, pulse rate 112x/minute, respiratory rate 20x/minute, and temperature 37.1°C. Physical examination of the head, neck, and chest showed no abnormalities; the abdomen was scaphoid in shape, tympanic with presence of bowel peristaltic sounds, epigastric tenderness on palpation. The working diagnosis of the patient was

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achalasia with the differential diagnosis of pseudoachalasia (caused by malignancy).

Esophagography demonstrated a narrowing of esophagus lumen distal to the esophagogastric junction with a rat tail sign (**Figure**).

The patient was diagnosed with achalasia. The management was laparotomy with Heller myotomy and fundoplication.

The patient was allowed to have oral fluid intake two days after surgery; and discharged after the patient was able to eat and drink orally. She was advised towards taking a thicker and a more solid form of food diet, and to regularly replace the wound dressing as well as to keep clean. At 1-week follow-up, the patient's condition improved, she was able to eat porridge and began to gain weight. At 1-month follow-up, the patient was able to eat solid food, and gained more weight.

DISCUSSION

Achalasia is a primary esophageal motility disorder characterized by the absence of esophageal peristalsis and impaired lower esophageal sphincter (LES) relaxation in response to swallowing.[5] LES pressure is found to be high in about 50% patients.[6] This condition cause a functional obstruction at the gastroesophageal junction.[1]

Achalasia was first described by Sir Thomas Willis in 1672. In 1881, Mikulicz described the condition as a cardiospasm and demonstrate 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.[7]

Achalasia is very rare, with an annual incidence of approximately 1 per 100,000 and a prevalence of 10 per 100,000 with no age, race, or gender predominance.[3] For some unknown reason, the incidence of achalasia increases in individuals with spinal cord injury, typically related to cervical and thoracic vertebral damage.[8] Altered esophageal motility is sometimes seen in patients with anorexia nervosa.[9] It is also seen in patients following eradication of esophageal varices by endoscopic sclerotherapy, in association with an increased number of endoscopic sessions but not with manometric parameters. Features of esophageal motility after endoscopic sclerotherapy are a defective lower sphincter and defective and hypotensive peristalsis.[10] Achalasia occurs with equal frequency in both males and females.

This disorder is mainly found in adult between the second to the fifth decade of life, with a peak incidence between the ages of 30 to 60[3]; it is very rare among children, less than 2% to 5% cases occur in children less than age 16. The mean incidence in children is 0.1 per 100,000 per year.[11] This case was 16 years old while her symptoms first started at the age of 10.

Achalasia is thought to be associated with the degeneration of the myenteric plexus and vagus nerve fibers to the lower esophageal sphincter, causing a loss of inhibitory neurons containing vasoactive intestinal peptide (VIP) and nitric oxide synthase at the esophageal myenteric plexus, and in severe cases, also involves cholinergic neurons. The exact cause of this degeneration is not clearly elucidated though many theories have been proposed, including autoimmune mechanism, viral infection, and genetic predisposition.[12] A European study compared immune-related deoxyribonucleic acid (DNA) in persons with achalasia with controls and found 33 single-nucleotide polymorphisms (SNPs) associated with achalasia. All of them were found in the major histocompatibility complex (MHC) region of chromosome 6, a location associated with autoimmune disorders such as multiple sclerosis, lupus, and type 1 diabetes.[13]

The esophagus is the conduit of transport for food bolus from mouth to stomach, and also prevents reflux of the stomach contents. This function achieved through coordinated peristaltic contractions in the pharynx and esophagus pared with the relaxation of the upper and lower esophageal sphincters.[1] LES pressure and relaxation are regulated by excitatory (eg, acetylcholine, substance P) and inhibitory (eg, nitric oxide, vasoactive intestinal peptide) neurotransmitters.[14] Persons with achalasia lack non-adrenergic, non-cholinergic, inhibitory ganglion cells, causing an imbalance in excitatory and inhibitory neurotransmission; the result is a hypertensive non-relaxed esophageal sphincter.[12] Gradual neural degeneration directly causes excessive contractions of LES and a loss of regulation, leading to functional obstruction which subsequently results in dilatation. This dilatation results in an irreversible aperistalsis

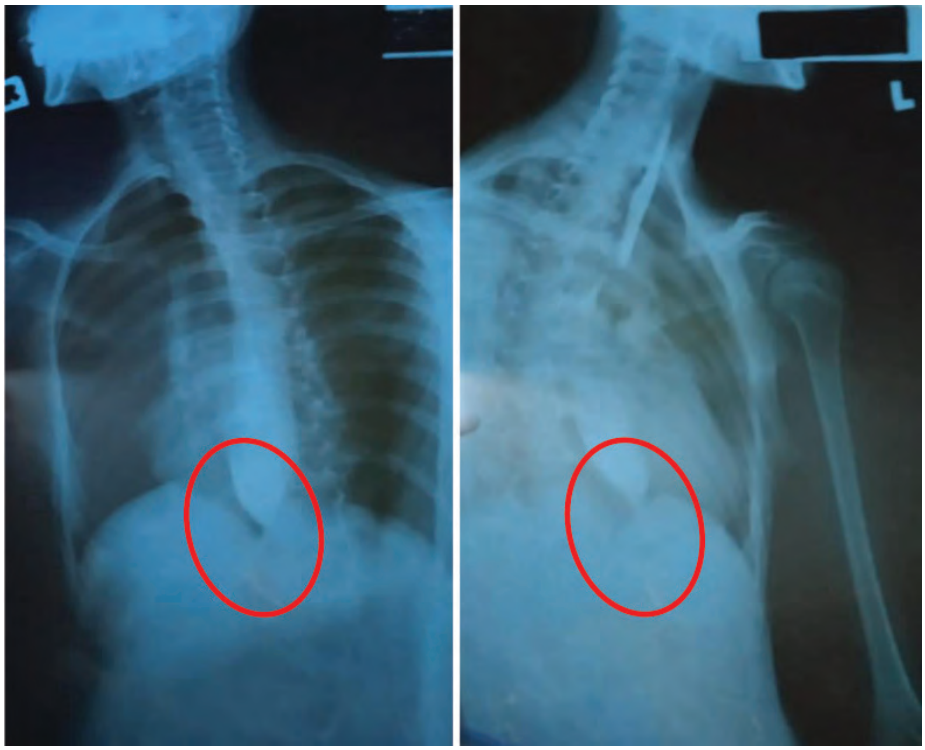

Figure. Esophagogram showing rat tail sign.


and worsening obstructive symptoms. The reason that these changes occur is unclear.[1] In this case, the obstructive symptoms worsened over months possibly due to these consecutive processes.

Most achalasia patients present with dysphagia, initially more severe on solids than on liquids though 70-97% patients will have dysphagia to both solids and liquids at presentation.[15] Regurgitation of undigested, retained food occurs in about 75% patients. More than half patients will present with chest pain, which must be differentiated from cardiac angina pectoris. About 60% achalasia patients may have some degree of weight loss at presentation due to poor esophageal emptying and decreased or modified food intake. Less common symptoms are hiccups or belching difficulty. The most common extraesophageal manifestations are pulmonary complications. Structural or functional pulmonary abnormalities occur in more than half patients, and might be due to recurrent aspiration or tracheal compression from a dilated esophagus.[16] Physical examination may reveal an emaciated individual.[1,17] Our case initially complained of dysphagia to both solids and liquids starting at the age of 10 years, and progressively worsened until she was not able to swallow any solids and liquids at 16 years old. At presentation, the patient's chief complaint was persistent vomiting of solids and liquids with same shape, consistency, and color, a sign of regurgitation of undigested food. She also complained of weight loss and general weakness supported by emaciated condition. An esophageal achalasia was suspected and diagnostic studies were planned.

Diagnostic studies to confirm the disease are needed in clinical suspicion for achalasia since symptoms are nonspecific. It is important to exclude benign and malignant causes of lower esophageal obstruction. The best initial test to diagnose achalasia is barium esophagogram (barium swallow); the classic finding is the smooth tapering of the lower esophagus to a "bird's beak" appearance, with dilatation of the proximal esophagus and lack of peristalsis during fluoroscopy. Some cases reveal an air-fluid level and absence of intra-gastric air while in advanced disease, a sigmoid-like appearance of the esophagus may be visible.[18] A timed barium swallow is used to assess esophageal emptying. This variant of classic barium swallow is performed by drinking 236 ml barium in upright position and radiographs were taken at one, two, and five minutes after the last swallow. The height of the barium column after five minutes, and the esophageal width are measured pre and post-treatment.[17] In this case, a barium esophagogram revealed a narrowing lumen of distal esophagus to the esophagogastric junction with a rat tail sign or "bird's beak" appearance (**Figure**), which supported the diagnosis of achalasia.

Upper endoscopy (esophagogastroduodenoscopy - EGD) is recommended for all patients with suspected achalasia or dysphagia to exclude premalignant or malignant esophagus lesions. EGD has low accuracy for diagnosis of achalasia and may be normal in the early stages of the disease.[19] Findings in advanced cases include rosette appearance of the esophagogastric junction or esophagus which has become dilated, tortuous, and atonic, often with retained food and saliva. The esophagus may be normal or show evidence of esophagitis due to chronic stasis. A firm resistance of the scope passing through the esophagogastric junction, especially in an older patient or case with a short duration of symptoms and significant weight loss, should raise concern for pseudoachalasia, especially malignancy.[20] Useful studies for pseudoachalasia are CT scan, endoscopic ultrasound (to rule out submucosal lesions), and transabdominal ultrasound.[17]

The most sensitive test and the gold standard for diagnosis of achalasia is esophageal manometry.[21] Manometry will demonstrate incomplete LES relaxation in response to swallowing, sometimes a lack of peristalsis in the lower esophagus, and a pressure increase in the lower esophageal sphincter.[22]

This case did not undergo EGD since we did not suspect any premalignant or malignant esophagus lesions. Esophageal tumor is rare in children and causes dysphagia for solids more than liquids. We did not peform manometry because the diagnostic tool was not available. We determined diagnosis of achalasia based on clinical and barium esophagogram findings.

Treatment is to decrease the outflow resistance caused by a non-relaxing and hypertensive lower esophageal sphincter. Current treatment modalities for primary idiopathic achalasia are nonsurgical or surgical. Nonsurgical options include pharmacotherapy, endoscopic botulinum toxin injection, or pneumatic dilatation. Surgical options are laparoscopic Heller myotomy (LHM) and peroral endoscopic myotomy (POEM). Pharmacologic treatments include the administration of nitrates, calcium channel blockers, and phosphodiesterase-5 inhibitors to reduce lower esophageal sphincter (LES) pressure.[23] These treatments are less effective, provide only short-term symptom relief, and are primarily reserved for patients who are waiting for or who refused more definitive therapy, such as pneumatic dilatation or surgery.[1,22]

For most achalasia patients, treatment with pneumatic dilation, surgical myotomy, or peroral endoscopic myotomy (POEM) is more recommended rather than botulinum toxin injection. Pneumatic balloon dilation and surgical myotomy have comparable high success rates. However, the efficacy of both treatments decreases over time and approximately one-third to one-half of patients will require repeat treatment within 10 years.[24] Although botulinum toxin has high initial success rates comparable to pneumatic dilation and surgery, patients treated with botulinum toxin have more frequent and shorter time to relapse.[25]

Laparoscopic Heller myotomy (LHM) with a partial fundoplication is the optimal surgical treatment of achalasia, with effective control of symptoms in 90 to 97 percent of patients.[26] This procedure will cut the circular muscle fibers running across the lower esophageal sphincter, leading to relaxation. LHM can potentially cause uncontrolled gastroesophageal reflux, so it typically pairs with an anti-reflux procedure such as Nissen, the posterior (Toupet), or the anterior (Dor) partial fundoplication. Laparoscopic Heller myotomy (LHM) with partial fundoplication is the surgical procedure of choice because complete (Nissen) fundoplication can cause severe postoperative dysphagia. Toupet partial fundoplication (270° posterior wrap of the fundus around the esophagus) reduces the risk of recurrent dysphagia by splaying open the edges of myotomy, thus preventing scarring, while Dor fundoplication (180° anterior wrap) provides coverage of the

primary repair when there is an esophageal perforation. The clinical success rate of LHM is high (76 to 100%) at 35 months, with a low mortality rate of 0.1%.[27] Disease progression after five years subsequently reduces the success rate.[22] An open approach (laparotomy) to the Heller myotomy is rarely performed as the initial treatment and is reserved for patients who have had multiple prior abdominal operations or who cannot tolerate a pneumoperitoneum because of cardiac or pulmonary disease.[25] In our patient, we performed an open (laparotomy) surgical myotomy with fundoplication.

Peroral endoscopic myotomy (POEM) is an effective minimally invasive alternative to laparoscopic Heller myotomy at limited centers. Dissection of the circular fibers of the LES is achieved endoscopically, leading to relaxation of the LES; however, the risk of gastroesophageal reflux is high because it does not include an anti-reflux procedure.[28]

The most common complications following laparoscopic and open myotomy with fundoplication include perforation, recurrent dysphagia, and gastroesophageal reflux.[29] Following laparoscopic Heller myotomy, the morbidity rate ranges between 1 and 10 percent and the mortality rate is <0.1 percent in the 30 day perioperative period.[25]

The principle components of postoperative care for laparoscopic and/or open myotomy include diet and control of nausea. Clear liquids are typically started following laparoscopic surgery, or after bowel function return following an open procedure. If no dysphagia occurs, the diet is advanced to a soft diet in the following day. If there is episodes of dysphagia, patients are maintained on a full liquid diet for a longer period. Aggressive treatment with antiemetics is provided to avoid emesis, which can lead to rupture of the repair. Medications can be administered in a crushed form until the patient is tolerating a regular diet. In patients undergoing Heller myotomy with partial fundoplication, antacids or proton pump inhibitors are used only if patients experience symptoms of heartburn and regurgitation and a pH monitoring study reveals gastroesophageal reflux. Antacids or proton pump inhibitors are typically recommended for patients undergoing a Heller myotomy alone.[25] In our patient, the advancement of diet went smoothly without any postoperative complications. At 1-month post-surgery, she was able to eat solid foods and gain weight.

CONCLUSION

Esophageal achalasia is rarely found in pediatric population. High clinical suspicion is required as history often provide key elements for diagnosis. In low resource settings, barium esophagogram is often the only available diagnostic study to confirm the diagnosis. Heller myotomy plus an anti-reflux procedure (fundoplication), whether performed laparoscopically or with an open (laparotomy) approach, is the standard of treatment in children. The principle component of postoperative care is advancement of diet, control of nausea, and monitoring of post-operative complications.

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