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CASE REPORT

ACHALASIA OF THE CARDIA - A PEDIATRIC CASE REPORT

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ABSTRACT

Introduction: Achalasia is an esophageal motor disorder characterized by aperistalsis of the esophageal body and lack of relaxation of the lower sphincter in response to swallows. It affects both sexes and all age groups. The exact etiology of this degeneration is unclear though many theories have been proposed. Possible etiology of achalasia can be familial, infection or autoimmune. Patients often present with progressive dysphagia to solids and liquids, heartburn, chest pain, regurgitation, and varying degrees of weight loss or nutritional deficiencies.

Case report: In January 2020, a 10 years old boy was admitted to our hospital due to vomiting, chest pain during swallowing, and prolonged cough. A lot of investigations were performed. Fyberbronchoscopy showed that the trachea, the right main bronchus and the left main bronchus were with anteroposterior narrowed lumen. Computed tomography of thorax and abdomen showed dilated esophagus with largest diameter of 45 mm subcarinal with retained content.There is a compression of the right hillus. The finding indicates achalasia of the cardia. One month after the hospital stay, the child was admitted at the Clinic for pediatric surgery. Laparoscopic Heller Myotomy (LHM) with antireflux Dor fundoplication was performed. The following period the boy has no gastric or respiratory symptoms.

Conclusion: We can conclude that it is worth to suspect achalasia in a pediatric patient with prolonged cough and recurrent respiratory infections refractory to conventional treatments such as bronchodilators, especially if the patient also has a history of vomiting and dysphagia.

Keywords: achalasia, esophagus, pediatric

INTRODUCTION

Achalasia is an esophageal motor disorder characterized by aperistalsis of the esophageal body and lack of relaxation of the lower sphincter in response to swallows. It affects both sexes and all age groups [1,2]. Achalasia was first described by Willis [3] in 1674 as “food blockage in esophagus”. He treated these patients successfully with a dilator made of whale bone and sponge [3].

Achalasia (AC) is a major primary esophageal motor disorder with a reported incidence of approximately 1.63/100,000 of population [4]. Recent studies have shown that the actual incidence is far higher than that previously reported [5,6].

Most of cases are idiopathic, but the syndrome can be associated with malignancy (especially involving the gastroesophageal junction) and as a part of the spectrum of Chagas disease. Rarely, achalasia is genetically transmitted [7].

Achalasia is thought to occur from the degeneration of the myenteric plexus and vagus nerve fibers of the lower esophageal sphincter. [8,9] There is a loss of inhibitory neurons containing vasoactive intestinal peptide (VIP) and nitric oxide synthase at the esophageal myenteric plexus, but in severe cases, it also involves cholinergic neurons [10,11].

The exact etiology of this degeneration is unclear though many theories have been proposed. Possible etiology of achalasia can be familial, infection or autoimmune.

The existence of familial cases may suggest that in some achalasia is an inherited disease [12-15]. Such familial cases have been mostly seen in the pediatric population, between siblings and in a few cases in monozygotic twins [12,13], and there are also a few reports of a parent-child association for achalasia [14].

Several studies, where measles and varicella zoster virus antibodies were found to be higher among a number of achalasia patients, have suggested a possible association between viral infections and achalasia [16,17]. Strong piece of evidence in favor of infection in the pathogenesis of achalasia, however, is the fact that Chagas disease, caused by Trypanosoma cruzi, very closely mimics the pathophysiology of primary achalasia [18].

An autoimmune etiology for achalasia has been considered because of the presence of neural inflammation in absence of conclusive evidence of infection. Studies have demonstrated inflammatory cell infiltrate of the myenteric plexus in 90%-100% of esophageal specimens from achalasia patients [19,20]. All of these evidences are not sufficient to conclude what is the etiology of achalasia. More studies are needed to explore the exact cause of this enigmatic disease.

Patients often present with progressive dysphagia to solids and liquids, heartburn, chest pain, regurgitation, and varying degrees of weight loss or nutritional deficiencies [21,22]. Classically, achalasia presents as progressive dysphagia to solids and liquids. Heartburn may present in 27% to 42% of patients with achalasia, and thus, patients are frequently misdiagnosed with gastroesophageal reflux disease (GERD) and treated with proton pump inhibitor (PPI) therapy [23] Younger patients are more likely to have chest pain and heartburn. Some achalasia patients may also experience respiratory symptoms such as cough, wheezing, and hoarseness. Respiratory symptoms are observed in over 40% of patients with achalasia. Most of these occur at least daily and may be secondary to retention of food with regurgitation, the mass effect of a dilated esophagus, or both [24].

CASE REPORT:

In January 2020, a 10 year old boy was admitted to our hospital due to vomiting, chest pain during swallowing, and prolonged cough.

From the anamnestic data, the child was healthy until 8 years of age, when he started to vomit after every meal, and lost 6 kg of his weight. He also had respiratory symptoms such as cough and wheezing, and was investigated for asthma, with positive skin prick tests for *Dermatophagoides*. He started taking inhaled topical corticosteroid fluticasone several months before admission to hospital. Because of the vomiting the patient was first sent at the University pediatric clinic at the gastroenterohepatology department. Many investigations, such as abdominal ultrasound, tests for celiac disease, coproculture, testing for Helicobacter pylori were made, and all of them were with normal finding. Two months ago, he was treated with Azythromycin because of *Mycoplasma pneumoniae* infection.

At admission, the boy was afebrile, pale, with frequent productive cough, moderate dyspnea, and auscultatory on the lungs with pneumonic finding and moderate bronchoopstruction. Heart sounds were rhythmic. Heart rate was 70 beats per minute. Arterial blood pressure was 100/70 mmHg. Palpation revealed that abdomen was soft and painless in all areas.

Diagnostic findings: Complete blood count – hemoglobin 140g/L, erythrocytes $4,5 \times 10^{12}$ leukocytes $7,9 \times 10^{9}$, Platelets 207×10^{9}, ESR 20/50 mm/h, CRP 2,5 mg/l.

Blood biochemistry: total protein 60 g/L, albumin 38 g/L, Na 139 mmol/l, K 4,07 mmol/l Cl 107 mmol/l, Hepatal enzymes: AST 21 u/l, ALT 19 u/l, blirubin 14 mcmol/l, gamma GT 8 u/l.

Urine- all findings were within normal range. Serology for *Helicobacter pylori* was negative.

Because of the prolonged cough investigations for tuberculosis were realized - Mantoux test, Quantiferon TB gold test, ARB and Lowenstein-Jensen culture, all of them were normal.

Chest X-ray showed accentuated lung pattern on the right side, without parenchymal consolidation (Figure 1).

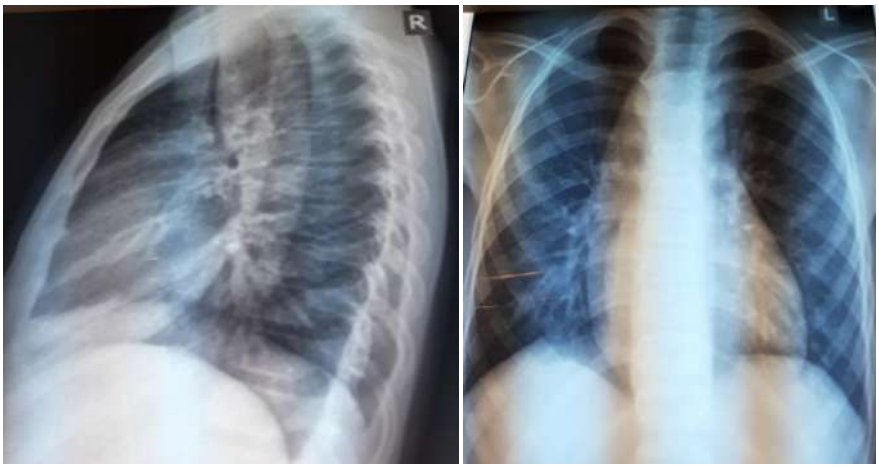

Fig. 1. Chest X-ray

Abdominal ultrasound was with normal findings.

Fyberbronchoscopy showed that the trachea, the right main bronchus and the left main bronchus were with anteroposterior narrowed lumen. The finding suggested malformation of the tracheobronchial tree with compression from the outside.

Because of this finding, computed tomography of thorax and abdomen was performed. It showed dilated esophagus with largest diameter of 45 mm subcarinal with retained content. There is a compression of the right hillus. The finding indicates achalasia of the cardia. (Figure 2)

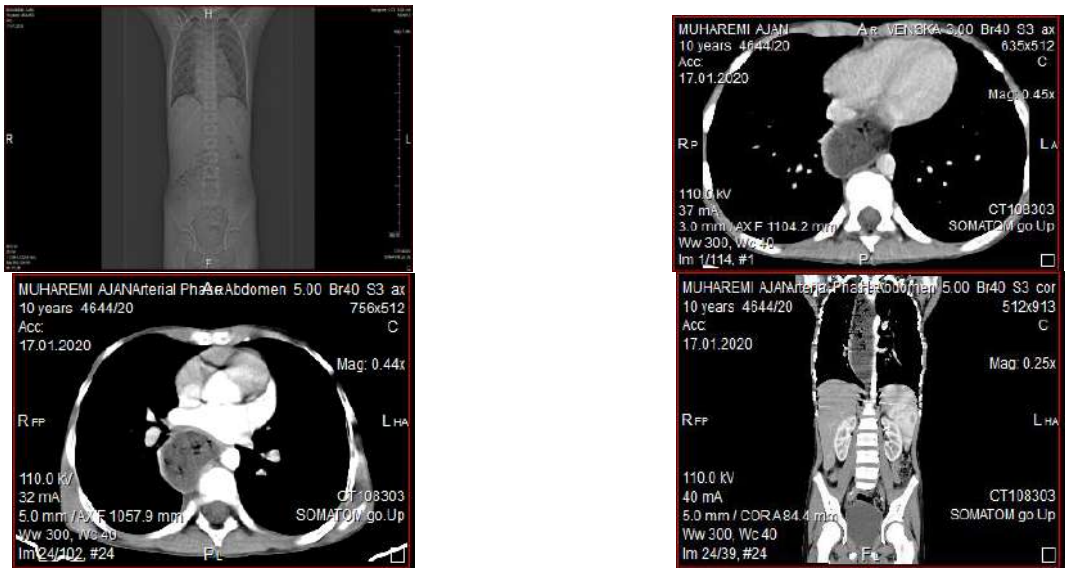

Fig. 2. CT of thorax and abdomen

The patient was treated with antibiotics, inhaled bronchodilator, and corticosteroids in the first several days, proton-pump inhibitor. During the hospital stay he had gastric pain and vomiting after meal in the first week, and the symptoms withdrew after we started the therapy with omeprazole and diet with small but more frequent meals. The auscultatory lung finding was getting better every day, until it was completely withdrawn. He was discharged from hospital with proton pump inhibitor therapy, inhaled corticosteroid Fluticasone, and with advice for diet. One month after the hospital stay, esophagogram with act of swallowing was performed, the diagnosis of achalasia was confirmed, and the child was admitted at the Clinic for pediatric surgery. Laparoscopic Heller Myotomy (LHM) with anti-reflux Dor fundoplication was performed. In the postoperative period the child was in good condition, with antibiotic and analgetic therapy, and after 2 weeks he was discharged from hospital. We were following the child's condition after the surgical treatment. In this period, he had no gastric or respiratory symptoms.

DISCUSSION:

This case report represents a 10 years old boy with idiopathic achalasia of the cardia, who had gastric symptoms and prolonged cough. The findings of tracheal and bronchial compression on fyberbronchoscopy leaded us to further investigations and finally to the diagnosis of achalasia.

Although respiratory symptoms are unlikely to precede the more typical symptoms of achalasia such as dysphagia and regurgitation, cough as a primary presenting symptom has been reported in the pediatric population [25].

Laparoscopic Heller Myotomy **(**LHM) performed with or without antireflux fundoplication (Dor) is a highly effective treatment for achalasia. Originally performed as an open thoracotomy and laparotomy, the less-invasive laparoscopic approach has similar efficacy rates but decreased morbidity [26,27]. Patients also need to understand necessary lifestyle changes following myotomy, such as the need to eat small food boluses in an upright position, which allows gravity to assist with food transit and never to lay flat but rather at 30 to 45 degrees due to increased risk for aspiration.

CONCLUSION: From this case report we can conclude that it is worth to suspect achalasia in a pediatric patient with prolonged cough and recurrent respiratory infections refractory to conventional treatments such as bronchodilators, especially if the patient also has a history of vomiting and dysphagia.

REFERENCES:

1. Ghoshal UC, Kumar S, Saraswat VA, Aggarwal R, Misra A, Choudhuri G. Long-term follow-up after pneumatic dilation for achalasia cardia: factors associated with treatment failure and recurrence. Am J Gastroenterol. 2004;99:2304-2310.
2. Francis DL, Katzka DA. Achalasia: update on the disease and its treatment. Gastroenterology. 2010;139:369–374.
3. Willis T. Pharmaceutice rationalis sive diatribe de medicamentorum operationibus in humano corpore. London: Hagae Comitis, 1674.
4. Sadowski DC, Ackah F, Jiang B, Svenson LW. Achalasia incidence, prevalence and survival. A population-based study. Neurogastroenterol Motil. 2010;22:e256–61.
5. Duffield JA, Hamer PW, Heddle R, Holloway RH, Myers JC, Thompson SK. Incidence of Achalasia in South Australia Based on Esophageal Manometry Findings. Clin Gastroenterol Hepatol. 2017;15:360–365.

6. Samo S, Carlson DA, Gregory DL, Gawel SH, Pandolfino JE, Kahrilas PJ. Incidence and Prevalence of Achalasia in Central Chicago, 2004- 2014, Since the Widespread Use of High-Resolution Manometry. Clin Gastroenterol Hepatol. 2017;15:366–373.
7. Gockel HR, Schumacher J, Gockel I, Lang H, Haaf T, Nöthen MM. Achalasia: will genetic studies provide insights? Hum Genet. 2010;128:353–364.
8. Cassela RR et al. Fine-structure changes in achalasia of esophagus. II. Esophageal smooth muscle. Am J Pathol. 1965;46:467–75.
9. Cassela RR, et al. Fine-structure changes in achalasia of esophagus I. Vagus nerves. Am J Pathol. 1965;46:279–88.
10. Francis DL, Katzka DA. Achalasia: update on the disease and its treatment. Gastroenterology. 2010;139(2):369–74.
11. Misiewicz JJ, Waller SL, Anthony PP, Gummer JW. Achalasia of the cardia: pharmacology and histopathology of isolated cardiac sphincteric muscle from patients with and without achalasia. Q J Med. 1969;38(149):17–30.
12. Frieling T et al. Family occurrence of achalasia and diffuse spasm of the esophagus. Gut. 1988;29:1595–1602.
13. Stein DT, Knauer CM. Achalasia in monozygotic twins. Dig Dis Sci 1982; 27:636-640.
14. Annese V, Napolitano G, Minervini MM, et al. Family occurrence of achalasia. J Clin Gastroenterol. 1995;20:329–330.
15. Bosher LP, Shaw A. Achalasia in siblings: clinical and genetic aspects. Am J Dis Child. 1981;135:709–710.
16. Jones DB, Mayberry JF, Rhodes J, Munro J. Preliminary report of an association between measles virus and achalasia. J Clin Pathol. 1983;36:655–657.
17. Robertson CS, Martin BA, Atkinson M. Varicella-zoster virus DNA in the esophageal myenteric plexus in achalasia. Gut. 1993;34:299–302.
18. de Oliveira RB, Rezende Filho J, Dantas RO, Iazigi N. The spectrum of esophageal motor disorders in Chagas' disease. Am J Gastroenterol. 1995;90:1119–1124.
19. Goldblum JR, Rice TW, Richter JE. Histopathologic features in esophagomyotomy specimens from patients with achalasia. Gastroenterology. 1996;111:648–654.
20. Raymond L, Lach B, Shamji FM. Inflammatory aetiology of primary esophageal achalasia: an immunohistochemical and ultrastructural study of Auerbach's plexus. Histopathology. 1999;35:445–453.
21. Vaezi MF, Pandolfino JE, Vela MF. ACG clinical guideline: diagnosis and management of achalasia. Am J Gastroenterol. 2013;108(8):1238–1249.
22. Francis DL, Katzka DA. Achalasia: update on the disease and its treatment. Gastroenterology. 2010;139(2):369–374.
23. Spechler SJ, Souza RF, Rosenberg SJ, Ruben RA, Goyal RK. Heartburn in patients with achalasia. Gut. 1995;37(3):305–308.
24. H. Sinan, R. P. Tatum, R. V. Soares, A. V. Martin, C. A. Pellegrini, B. K. Oelschlager, Prevalence of respiratory symptoms in patients with achalasia. Diseases of the Esophagus. 2011;24(4):224–228.
25. Kugelman A, Berkowitz D, Best L A, Bentur L. Upper airway obstruction as a presenting sign of achalasia in childhood. Acta Paediatr. 2000;89:356–8.
26. Vaezi MF, Richter JE. Current therapies for achalasia: comparison and efficacy. J Clin Gastroenterol. 1998;27(1):21–35.
27. Ali A, Pellegrini CA. Laparoscopic myotomy: technique and efficacy in treating achalasia. Gastrointest Endosc Clin N Am. 2001;11(2):347–358.