

CASE REPORT / ПРИКАЗ БОЛЕСНИКА

Esophageal achalasia in a two-year-old boy

Đorđe Savić^{1,2}, Maja Miličković^{1,2}, Predrag Ilić^{1,2}, Miroslav Vukadin¹, Dejan Stojakov^{2,3}¹Dr Vukan Čupić Mother and Child Health Care Institute of Serbia, Belgrade, Serbia;²University of Belgrade, Faculty of Medicine, Belgrade, Serbia;³Dr Dragiša Mišović – Dedinje University Clinical Hospital Center, Belgrade, Serbia**SUMMARY**

Introduction Esophageal achalasia is a neurodegenerative motility disorder, which is characterized by ineffective or absent esophageal peristalsis and the lack of hypertonic lower esophageal sphincter relaxation. Achalasia causes failure to thrive in children and can have serious respiratory complications. Achalasia is a very rare condition in pediatric population, and usually misdiagnosed as gastroesophageal reflux. The treatment of choice is Heller esophagocardiomyotomy.

The aim of this paper is to present a rare case of a two-year-old child with achalasia, diagnostic procedures, and successful operative treatment.

Case outline The patient's problems started at the age of six months, with audible breathing and respiratory stridor. The child was admitted at a local hospital at seven months of age, dismissed with dietary advices, again admitted at the age of 19 months, and transferred to our institution. Upper gastrointestinal series and computed tomography revealed findings characteristic for achalasia, and on esophagoscopy exam there was no opening of lower esophageal sphincter and cardia on insufflation. Pneumatic dilation was performed with temporary improvement. Laparotomic Heller esophagocardiomyotomy with Dor partial fundoplication was successfully performed.

Conclusion Achalasia is a very rare condition in infants and young children. There is often a delay when establishing the correct diagnosis. Upper gastrointestinal series and endoscopic exam are most reliable methods to detect achalasia. Pharmacological treatment, intrasphincteric injection of botulinum toxin and pneumatic dilations are not efficient methods, especially in small children. The method of choice in the treatment of achalasia is Heller esophagocardiomyotomy with partial fundoplication.

Keywords: achalasia; children; surgery

INTRODUCTION

Esophageal achalasia is a neurodegenerative motility disorder of the esophagus, characterized by ineffective esophageal peristalsis and the absence of relaxation of hypertonic lower esophageal sphincter (LES), due to degenerative changes of inhibitory fibers and ganglion cells of esophageal Auerbach myenteric plexus [1, 2]. The clinical symptoms of achalasia were first described by Thomas Willis in 1674. The term achalasia was introduced by A. F. Hertz in 1914 and Hurst in 1927. The term cardio-spasm has also been used for a long time. Some authors believe that achalasia could arise due to the disorder of extraesophageal innervation via the dorsal nuclei of the vagal nerve [2]. Cassella showed that in 68% of esophageal samples of patients with achalasia, the degeneration, or absence of ganglion cells were present [3].

The incidence of achalasia is 1:100,000 people, with symptoms mainly occurring in adolescents and adults between the ages of 30 and 60 [1, 4, 5]. Only 3–5% of patients are younger than 14 years, and the disease is extremely rare in infancy [5]. However, a case of premature infant weighing 1200 gr with achalasia is reported [5]. Some authors found the annual incidence of achalasia in children 0.11:100,000, mostly between the ages of seven and 12 [1]. Achalasia

can be a primary congenital disorder, a consequence of some infectious diseases (Chagas disease, varicella and viral esophagitis), or autoimmune diseases (Guillain–Barre acute polyradiculoneuritis, Sjögren syndrome, eosinophilic esophagitis and scleroderma) [2]. Achalasia can be a part of Allgrove syndrome, together with alacrima and adrenal insufficiency (triple A) [4]. Some authors reported cases of transitory achalasia in infants with low birth weight or with Pierre Robin malformation [4]. Achalasia causes dysphagia, regurgitation, vomiting, aspiration of esophageal content with respiratory problems, anorexia, and failure to thrive. This disease can be complicated by obstructive bronchiolitis, lung abscess, pleural empyema, and pulmonary fibrosis.

The diagnosis of achalasia is established by esophageal manometry [6], esophagoscopy, upper gastrointestinal (GI) series and computed tomography (CT). The treatment can be conservative or surgical. Conservative treatment consists of lowering LES pressure pharmacologically (anticholinergic and antispasmodic medicaments) or by intrasphincteric injection of botulinum toxin. Nowadays, the conservative treatment can be applied only in very mild to moderate forms of achalasia, usually with unsatisfactory long-term effects [4]. Pneumatic dilation (PD) of the LES also can give only temporary

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positive results. The surgical treatment consists of Heller esophagocardiomyotomy (HE). The longitudinal splitting of smooth muscular fibers is made in the distal esophagus, cardia, and gastric fundus. An antireflux procedure should be performed at the same time, preventing postoperative gastroesophageal reflux (GER). Nowadays, HE is considered the treatment of choice for achalasia, especially in children.

CASE REPORT

The patient's problems started at the age of six months, when the intake of mushy and firmer foods began. Anamnestic data suggested that liquids and milk intake was pretty well tolerated. After food intake there was audible breathing, wheezing and respiratory stridor, without cyanosis. The child was admitted to a local hospital at seven months of age, lung radiographs, laboratory analyzes and otorhinolaryngological (ORL) examination were performed, and the patient was dismissed home with medical advice concerning food intake. At the age of 19 months the child was admitted again, and then transferred to our hospital. On admission, the child was malnourished, with body weight of 10.7 kg and body height of 83 cm. Lung auscultation revealed inspiratory stridor. Early psychomotoric development was regular. The patient's father was operated on because of hypertrophic pyloric stenosis. Pulmonary radiograph exam showed hilifugal bilateral salient interstitial drawing. Upper GI series revealed the markedly dilated esophagus, while its terminal part tapered conically and contrast, in a thin jet and in small amounts, passed into the stomach (Figure 1). Endoscopic exam revealed remained food in esophageal lumen, the dilated esophagus, and stenotic cardia not opening to insufflation. Esophageal narrowing at the level of the diaphragm was dilated by esophageal balloon N°10–12 to 2/3 of balloon width. For the first two weeks after dilation, the child's condition improved, but the recurrence occurred. The child was admitted again after 50 days, with the same complaints. The planned esophageal manometry at another institution could not be done because of the child's age. Chest CT scan recorded the dilation of the cervical and thoracic parts of the esophagus, with a maximum diameter of 17 mm in the anteroposterior (AP) direction, and 27 mm in the laterolateral (LL) direction. The esophageal wall was uniformly 3 mm thick, and hydroaeric level in the lower esophagus was prominent. When the patient was two years old, he was operated on. After medial laparotomy, the thoracic esophagus was mobilized, the vagal nerve identified and trapped. The longitudinal myotomy was performed, engaging 3 cm of the distal esophagus, cardia and 3 cm of the gastric fundus (Figure 2). The cardiomyotomy was difficult to perform due to the scar tissue, caused by previous PD. Esophageal and gastric mucosal prolapse occurred. The Dor anterior gastric fundoplication in the length of 2.5 cm was performed. Postoperative course was uneventful. Control upper GI series showed no contrast extralumination, esophageal dilation was still present, but contrast passed into the stomach faster and



Figure 1. Preoperative radiography

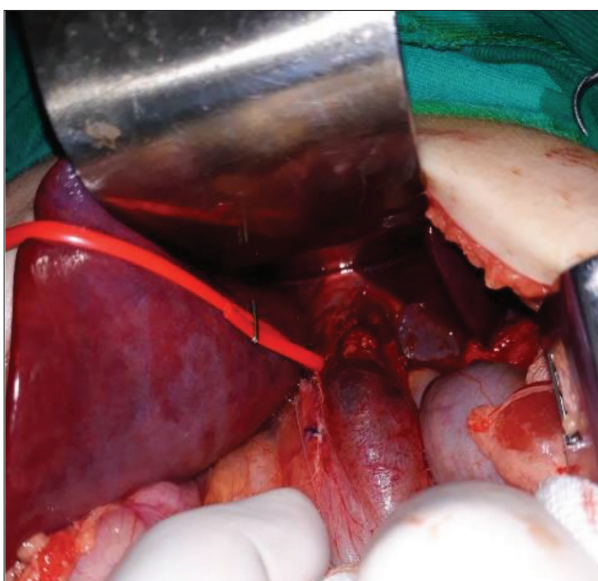


Figure 2. Operative myotomy

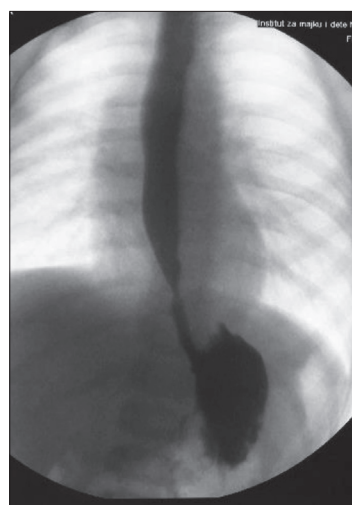


Figure 3. Postoperative radiography

in a thicker jet (Figure 3). The patient was readmitted two months later. Food intake was significantly better and major complaints reduced. Contrast upper GI study showed uniformly expanded esophagus, and the distal part of the esophagus moderately narrowed, but barium passed into the stomach without delay. There were no signs of GER.

Esophageal PD was performed and moderate stenosis at the cardia level completely dilated by the balloon 10–12 mm. After that, in six months follow-up the patient had no complaints or swallowing problems.

DISCUSSION

Esophageal achalasia in children is a rare disease and is often misdiagnosed as GER or, less commonly, as eating disorders, idiopathic failure to thrive, or bronchial asthma. Many studies have shown that achalasia in infants and young children manifests with difficulty feeding, nocturnal cough, wheezing, stridor, acute upper respiratory tract obstruction and recurrent respiratory infections, and in older children with symptoms of GER, sore throat, cough and hoarseness. Overall, the most presenting symptoms are regurgitation (83%), dysphagia (71%), poor growth (54%) and respiratory symptoms (41%) [7]. The diagnosis of achalasia is difficult to establish in children under five years of age, since its incidence is very low, and often there is a delay in making the diagnosis and applying the appropriate treatment [1]. Although 18% of pediatric patients have symptom onset during infancy, in only 6% of patients the diagnosis of achalasia is established during infancy [7]. In a meta-analysis made by Myers et al. [7], performed on 175 pediatric patients worldwide, achalasia has the predilection for male sex (61%:39%), and family cases and associated disorders are very rare. Poornachand et al. [1] state that the median time from symptoms onset to diagnosis is nearly three years. The patient's age and the rarity of achalasia in pediatric population are the reasons that our patient's condition was considered as distal esophageal peptic stenosis due to GER and treated by antiulcer therapy. Esophageal manometry is the gold standard, as it shows the elevated basal pressure of LES, the absence of its relaxation on swallowing, and the absence or reduction of esophageal peristalsis [1]. Unfortunately, this procedure was not performed in our patient because of his age. High-resolution impedance manometry can be very useful in the diagnostics [8, 9]. CT scan, contrast studies, and esophagoscopy are very reliable and useful diagnostic methods. Upper GI series, showing dilated, aperistaltic esophagus and narrowed esophagogastric junction as "bird beak" is diagnostic in more than 92% of patients [7]. Our patient's CT, radiography and esophagoscopy findings were characteristic.

The decreased esophageal peristalsis and the absence of LES relaxation cannot be repaired, so the treatment is aimed to reducing LES pressure. Conservative treatment of achalasia is based on the reduction of LES pressure by using anticholinergic and spasmolytic medications. Administration of isosorbide dinitrate, calcium channel blockers and nifedipine showed some positive effects in adults, but not in children [1, 8]. In our patient this kind of medical treatment was not applied because of his age. Intraspincteric injection of botulinum toxin also does not give satisfactory results [1]. The botulinum injection treatment was not considered as a therapeutic mode in our patient, considering negative results announced in many articles

and the child's age. In adult patients, PD can achieve success in 70–80% [10]. Azizkhan et al. [11], however, showed that in pediatric population only 25% of patients responded favorably to repeated PD, while 75% had to undergo a myotomy subsequently, emphasizing that PD was not successful in any child younger than nine years. Jung et al. [4] recorded the higher success rate of PD comparing to Heller operation (55.5%:44.5% after six months, and 65%:40% after 24 months), but they do not recommend PD in children below six years of age. PD of the esophagus was made in our patient because of the suspicion of distal esophageal peptic stenosis. Peroral endoscopic myotomy is a new method that is gradually gaining importance in the treatment of achalasia in children [1, 8, 12]. However, the surgical treatment, based on esophagocardiomyotomy, has proven to be the most effective method for the treatment of achalasia [1]. Esophagocardiomyotomy can be performed by an abdominal or thoracic approach, but the abdominal approach is preferable because of possibility of performing the antireflux procedure. Myotomy is followed by partial gastric fundoplication in order to reduce the incidence of postoperative GER (from 13% to 7%), although some authors consider it unnecessary [6]. Myotomy with fundoplication have higher success rate comparing to myotomy alone (91%:73%) [7]. The success rate of HE in children below six years of age is 75% at six months, and 83% at 24 months of follow up [2]. Some authors reported the overall success rate of the treatment of achalasia in children, by PD or HE, or both, of 57% after six months, and 64% after 24 months of follow up [7]. Other authors recorded success rate in children of 70–90% after 24 months, similar to the success rate in adults of 80–90% [4]. In our patient, the myotomy was made longitudinally and encompassed about 3 cm of the distal esophagus and 3 cm of the gastric fundus, with mucosal prolapse and without mucosal opening. The length of myotomy was shorter than presented in literature, considering the patient's age. Esophagocardiomyotomy was difficult, due to the scar tissue caused by previous PD. The Dor anterior gastric fundoplication in the length of 2.5 cm was performed in order to prevent postoperative GER, as well as to cover the denuded mucosa. Nowadays, laparoscopic esophagocardiomyotomy, with or without fundoplication, has gained primacy over laparotomic, as an effective method that allows quick recovery, short hospital stay and good definitive outcome [1, 13, 14, 15].

Despite a very low incidence in pediatric population, esophageal achalasia must be considered as a cause of regurgitation, failure to thrive and respiratory problems in infants and children. Once there is a suspicion, the diagnosis can be easily made by esophageal manometry, upper GI series endoscopic exam. The surgical treatment, consisted of HE and partial wrapping of gastric fundus, is a method of choice in the treatment of children with achalasia.

Informed consent statement: Consent was obtained from the patient's mother for the publication of this report and any accompanying images.

Conflict of interest: None declared.

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Ахалазија једњака код дечака од две године

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САЖЕТАК

Увод Езофагеална ахалазија је неуродегенеративни поремећај покретљивости једњака, који се карактерише неефикасном или одсутном перисталтиком једњака, као и одсуством релаксације хипертоничног доњег езофагусног сфинктера. Ахалазија узрокује застој у напредовању деце и може имати озбиљне респираторне компликације. Ахалазија је веома ретко стање у дечјој популацији и обично се погрешно дијагностикује као гастроезофагусни рефлукс. Метода избора у лечењу је Хелерова езофагокардиомиотомија. Циљ овог рада је да се прикаже редак случај ахалазије код двогодишњег детета, дијагностички поступци и успешно оперативно лечење.

Приказ болесника Тегобе су почеле у узрасту од шест месеци у виду чујног дисања и респираторног стридора. Болесник је примљен у локалну болницу у узрасту од седам месеци, отпуштен са препорукама о исхрани, а поново примљен у узрасту од 19 месеци и преведен у нашу установу

у. Контрастна радиографија и компјутеризована томографија су показали налазе карактеристичне за ахалазију, а на езофагоскопском прегледу није било отварања доњег езофагусног сфинктера и кардије при инсуфлацији. Учињена је пнеуматска дилатација са привременим побољшањем. Затим је лапаротомиски успешно учињена Хелерова кардиомиотомија са делимичном фундопликацијом по Дору. **Закључак** Ахалазија је веома ретко стање код одојчади и мале деце. Често постоји кашњење у постављању тачне дијагнозе. Контрастна радиографија и ендоскопски преглед су најпоузданије методе за постављање дијагнозе ахалазије. Фармаколошки третман, интрасфинктерична инјекција ботулиnum токсина и пнеуматска дилатација нису ефикасне методе, нарочито код мале деце. Метода избора у лечењу ахалазије је Хелерова езофагокардиомиотомија са делимичном фундопликацијом.

Кључне речи: ахалазија; деца; хирургија