## Case Report

# Achalasia Cardia in a 6-Month-Old Boy: Case Report and Review of Literature

Choudhury KM[a], Begum T[b]

**Abstract**

*Achalasia cardia is commonly seen in adult population. It is extremely rare in infants and seldom found in children. Diagnosis of the disease is difficult in children particularly in small infants because the typical symptoms are missing and hardly there are any sign.High index of suspicion should be exercised to bring it among the differentials.*

*We have described a 6-month-old boy with achalasia cardia and reviewed recent literatures to elucidate current scenario.*

***Key words:*** *achalasia cardia; children; infant.*

***Birdem Med J 2016; 6(1): 46-49***

### Introduction

Achalasia cardia is a primary motility disorder of the esophagus characterized by absence of effective peristalsis and a failure of relaxation of the lower esophageal sphincter (LES) in response to swallowing. The disease is most prevalent and commonly diagnosed in adults. It is occasionally seen in adolescent and late childhood but is rare in young children.[1] There are only a few cases with early infantile onset.[2] Less than 4% of all affected patients are symptomatic prior to age 15.[3]

The exact cause of achalasia cardia is not known. The absence of peristalsis is believed to be due to absence of ganglion cells or degeneration of myenteric nerve plexus of the esophagus. The LES fails to relax because of progressive disturbance in the intrinsic innervation due to reduced or absent nitric oxide synthase (NOS) activity.[4] The enzyme is responsible for synthesis of nitric oxide (NO) - a neurotransmitter of smooth muscle relaxation. Histochemistry also shows a significant reduction in other neurotransmitters namely vasoactive intestinal peptide, galanin and neuropeptide Y.[5,6]

The diagnosis of achalasia cardia with early onset is quite enigmatic. The pathognomonic symptom is dysphagia or retrosternal discomfort- which is purely a subjective sensation- can not be elicited from a non-verbal infant. Often the parents of the infant use the term vomiting instead of regurgitation to describe the expulsion of milk through the mouth. It may be confounding because regurgitation usually points to esophageal pathology whereas vomiting indicates stomach or more distal gastrointestinal tract (GIT). Failure to gain weight or gradual loss of weight and repeated respiratory infection with nocturnal cough are nonspecific though relevant in the diagnosis. Physical examination is often unrevealing. Endoscopy is not feasible in infants and esophageal manometry is not widely available. Barium swallow remains one of the most important tools for the diagnosis of achalasia.

### Case Report

A 6-month-old boy with vomiting since birth presented in an emaciated malnourished state. There was obvious failure to thrive (body weight was 4.3 kg, below 3rd percentile). Non-bilious, non-projectile vomiting used

**Author Informations**

a. Dr. Kamal M. Choudhury, MS (Pediatric Surgery), Associate Professor, Department of Pediatric Surgery, BIRDEM, Dhaka.

b. Dr. Tahmina Begum, FCPS (Pediatrics), Professor, Department of Pediatrics, BIRDEM, Dhaka.

**Address of correspondence:** Dr. Kamal M. Choudhury, Associate Professor and Head, Department of Pediatric Surgery, BIRDEM General Hospital and Ibrahim Medical College, 122, Kazi Nazrul Avenue, Shahbag, Dhaka-1000. E-mail: kamalemcee @gmail.com

**Received:** April 12, 2015 **Accepted:** November 30, 2015

to occur following most of the feeds. The boy suffered from respiratory tract infection (RTI) for three to four occasions since birth for which pediatric consultations were sought. Physical examination showed mild dehydration and pallor but there was no jaundice. Abdominal examination revealed no visible peristalsis or any mass in the abdomen. There was no organomegaly. His bowel and bladder habit were normal. Our provisional diagnosis was gastroesophageal reflux disease (GERD). Complete blood count (CBC), serum electrolytes, creatinine, blood sugar and liver function tests were within normal limit. Chest x-ray showed features of pneumonitis and absence of fundic gas shadow (Figure 1). Abdominal ultrasonography and contrast upper GIT series excluded hypertrophic pyloric

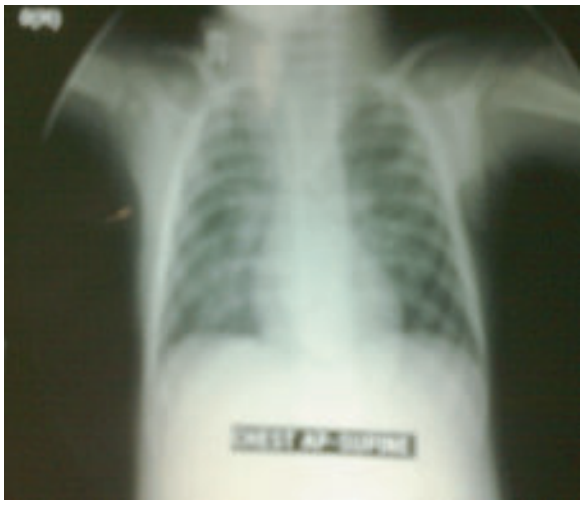

**Figure-1:** *X-ray chest shows absence of fundic gas shadow and features of pneumonitis.*

stenosis and other congenital anomalies of stomach and duodenum. The diagnosis of achalasia was made from absent fundic gas shadow in the chest x-ray and from the mega-esophagus with considerable retained contrast with smooth tapering of the esophagus in a pencil-tip or bird's beak pattern in the contrast upper GIT series (Figure 2). The patient was then prepared for Oesophago-cardiomyotomy. Following surgery, the boy did well; oral feeding was resumed on the 3rd post-operative day and there was no problem-namely vomiting or regurgitation related to feed. During follow-ups, the boy was thriving well with optimum weight gain. A contrast esophagogram in trendelenberg position showed no evidence of gastroesophageal reflux.

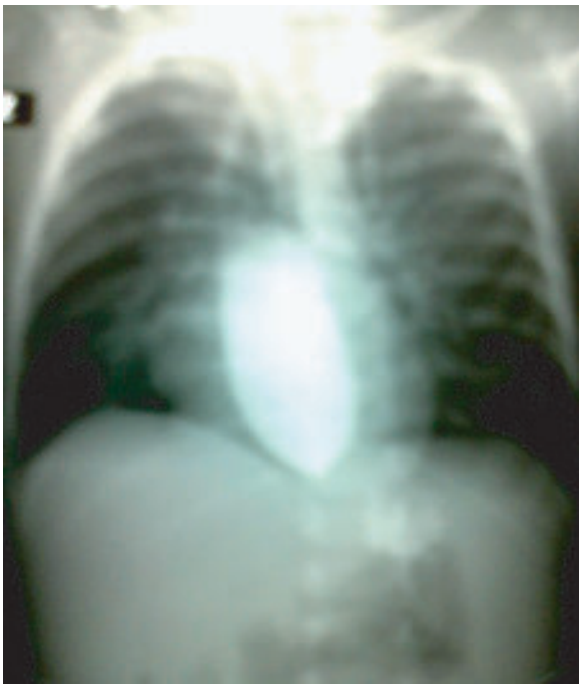

***Figure-2:*** *Contrast X-ray barium meal upper GIT showing mega-esophagus with retained contrast with smooth tapering at the distal end of esophagus (pencil-tip or bird's beak appearance)*

**Discussion**

We intend to report this case because achalasia is very uncommon in early infancy and also to underscore the difficulty in establishing a confirmed diagnosis in a small child. The problem starts right from history-taking. The patient can not tell about his complaints. The cardinal tell-tale symptom of achalasia is dysphagia with food "getting stuck" at some level in the chest. Only older children and adolescents will be able to describe symptoms in such detail. Moreover, parents insist that the act of expulsion was vomiting and not regurgitation as they fail to appreciate the difference between the two. However, patients with achalasia may actually vomit, particularly when the disease is advanced with huge mega-esophagus as, in such condition, the distal end of esophagus can actively contract to initiate vomiting. In a global survey of pediatric cases, Myers NA et al. reported a wide distribution of the age of onset of symptoms with only 18% developing symptoms during infancy. But, only in 6% cases a diagnosis of achalasia could be made.[7]

The reason why the symptoms of achalasia are not apparent in early childhood, is that it is a progressive

disease. The degeneration of intrinsic innervation of esophagus gradually gets worse as age progresses with obvious manifestation of the symptoms during adolescence and adult life. On the contrary, a few newborn, rarely, may develop regurgitation due to achalasia since birth, but for understandable reasons, it is under evaluated until the symptom persists long enough to affect the infant's growth and well-beings. In the present case, the boy developed vomiting during early neonatal period but much attention had not been paid. The vomiting persisted and the child suffered from repeated RTI for which symptomatic treatment was given. Still, vomiting persisted and there was failure to thrive. At that stage, at the age of 6 months, a delayed diagnosis of achalasia was made from contrast upper GIT series. A literature search had revealed that 31 cases of achalasia cardia in infants had been reported so far.[7] Among them, only in 3 patients the symptoms were apparent at neonatal stage. There was only one report of achalasia cardia in a premature neonate.[8] The preterm (29 weeks) baby was asymptomatic and the diagnosis was made by incidental x-ray chest done at 36 hours of age and confirmed by an esophagogram done at 2 weeks of age.

It is likely that early infantile achalasia may have a different etiology. Thomas RJ, et al. stated in their 10-year experience that achalasia in infants may be due to central cause (primary abnormality of dorsal vagal nucleus) unlike degeneration of ganglion cells of esophagus seen in older children and adults.[9] The degeneration is progressive and may be secondary to viral infection, Chagas disease or GERD.[10] Park and Vaezi also shared the view that primary neurogenic degeneration in the dorsal motor nuclei may be responsible for achalasia in infants.[11]

In our patient, the vomiting was present from birth- so suspicion of GERD, microgastria, pyloric web/atresia, gastric volvulus and duodenal (first part) web/atresia was logical. Hypertrophic pyloric stenosis was considered because of high prevalence though in this condition vomiting typically ensues during or after 2nd week of life. A contrast upper GIT series would be helpful in the diagnosis.

Differentials relating esophageal pathology included congenital esophageal stenosis, esophageal stricture due to GERD, diffuse esophageal spasm and nutcracker esophagus. The latter two are primary motility disorder of esophagus that usually present in adolescent and adults. The former two usually manifest during the early life. To confirm the diagnosis, one would need to resort to esophagoscopy and manometry.[12,13] In the present case, manometry could not be done due to unavailability. Endoscopic examination was also omitted considering infantile age of the boy. Fibre optic esophagoscopy is important in distinguishing achalasia from congenital esophageal stenosis or stricture as the instrument passes easily through the esophago-gastric junction in achalasia but can not be negotiated in stenosis or stricture. The differentiation between stenosis and stricture can be made by contrast upper GIT which shows smooth tapering of the lower end of the esophagus in the former whereas in the latter there are mucosal asymmetry and irregularity.

During surgical procedure, grossly dilated distal esophagus with thick hypertrophied muscle layer was identified and a modified Heller's esophagomyotomy was performed. The shallow (1-2 mm) linear incision extended about 5.5 cm on the distal esophagus and 0.5 cm onto the cardiac end of the stomach. Fundoplication was not done. Some surgeons profess an anti-reflux procedure to prevent complications of gastroesophageal reflux disease (GERD).[14-16] On the other hand, many authors prefer to omit anti-reflux procedure in these patients.[17,18] Clinically significant GERD is reported to occur in 15% to 20% of children following surgery.[19] Moreover, a relative esophageal obstruction may be created from the fundal wrap in the face of significantly impaired esophageal motility. With such consideration, a moderate approach is more appealing, with anti-reflux procedure reserved only for patients having symptomatic GERD following myotomy. In this regard, Ellis emphasized that limiting the distal extension of myotomy to within 1 cm from gastroesophageal junction could reduce GERD and esophagitis.[20]

After surgery, most of the patients are relieved of the symptoms and gain weight. A few children may continue to have minor problem and often require a few swallows of water during meals. Long-term follow-up is essential as the inherent pathology of esophageal dysmotility remains. Approximately 5% to 15% patients with achalasia may develop squamous cell carcinoma of esophagus after an average interval of about three decades.[21,22]

### *Conclusion*

Achalasia cardia is very rare in infants. Older children may present with dysphagia and regurgitation, but in the younger child the symptoms of this condition can be confused with those of GERD. A high index of suspicion must be borne in mind when encountered with an infant with repeated RTI, regurgitation and failure to thrive. The need to perform a concomitant fundoplication is controversial. It is better to do Heller's myotomy (with Ellis' modification) and reserve anti-reflux procedure for patients with evidence of symptomatic GER. Long-term follow-up is mandatory as there is increased risk of malignancy.

### References


1. Chatterjee S, Gajbhiye V, De A, Nath S. Achalasia Cardia in Infants: Reports of Two Cases. JIMA 2012; 44 Published 2013-Apr-10. DOI: http://dx.doi.org/10.5915/44-1-9260
2. Asch MJ, Liebman W, Lachman RS. Esophageal achalasia: diagnosis and cardiomyotomy in a newborn infant. J Pediatr Surg 1974; 9: 911-12.
3. Azizkhan RG, Tapper D, Eraklis A. Achalasia in childhood: A 20-year experience. J Pediatr Surg 1980;15: 452-56.
4. Takahashi T. Pathophysiological significance of neuronal nitric oxide synthase in the gastrointestinal tract. J Gastroenterol 2003; 38: 421-30.
5. Spitz L, Ure B. Achalasia. In: Spitz L, Coran AG (editors). Operative Pediatric Surgery.7thed. CRC Press, Taylor & Francis Group, UK; 2013, 354-67.
6. Khelif K, De Leat MH, Chaouachi B. Achalasia of the cardia in Allgrove's (triple A) syndrome: Histopathological study of 10 cases. Am J Surg Pathol 2003; 27:667-72.
7. Myers NA, Jolley SG, Taylor R. Achalasia of the cardia in children: a worldwide survey. J Pediatr Surg 1994; 29: 1375-79.
8. Shettihalli N, Venugopalan V, Ives NK, Lakhoo K. Achalasia cardia in a premature infant. BMJ Case Reropts;2010; 2010:bcr 0520103014; Published online 2010, November 5. doi: 10 1136/bcr.05.2010.3014.
9. Thomas RJ, Sen S, Zachariah N, Chacko J, Mammen KE. Achalasia cardia in infancy and childhood: an Indian experience. J R Coll Surg Edinb 1998; 43:103-4.
10. Upadhyaya VD, Gangopadhyaya AN, Gupta DK. Esophageal achalasia of unknown etiology in infants. World J Pediatr 2008; 4:63-65.
11. Park W, Vaezi MF. Etiology and pathogenesis of achalasia: the current understanding. Am J Gastroenterol 2005;100:1404-14.
12. Berquist WE, Byrne WJ, Ament ME. Achalasia: Diagnosis, management and clinical course in 16 children. Pediatrics 1983; 71: 798-805
13. Goldani HA, Staiano A, Borrelli O. Pediatric esophageal high-resulationmanometry: Utility of a standardized protocol and size-adjusted pressure topography parameters. Am J Gastroenterol 2009; 105: 460-67.
14. Vane DW, Cosby K, West K, Grosfeld JL. Late results following esophagomyotomy in children with achalasia. J Pediatr Surg 1988; 23: 515-19.
15. Morris-Stiff G, Khan R, Foster ME, Lari J. Long-term results of surgery for childhood achalasia. Ann R Coll Surg Engl 1997; 79: 432-34.
16. Rothenberg SS, Patrick DA, Bealer JF, Chang JH. Evaluation of minimally invasive approaches to achalasia in children. J Pediatr Surg 2001; 36: 808-10.
17. Pastor AC, Mills J, Marcon MA. A single center 26-year experience with treatment of esophageal achalasia: Is there an optimal method? J Pediatr Surg 2009; 44: 1349-54.
18. Vaos G, Demetriou L, Velaoras C, Skondras C. Evaluating long-term results of modified Heller limited esophagomyotomy in children with esophageal achalasia. J Pediatr Surg 2008; 43: 1262-69.
19. Nakayama DK, Shorter NA, Boyle JT. Pneumatic dilatation and operative treatment of achalasia in children. J Pediatr Surg 1987; 22: 619-22.
20. Ellis FH. Surgical management of esophageal motility disturbances. Am J Surg 1980; 139: 752-59.
21. Cohen SE. Motor disorders of esophagus. N Eng J Med 1979; 301:184-92.
22. Jaakkola A, Reinikainen P, Ovaska J, Isolauri J. Barrett's esophagus after cardiomyotomy for esophageal achalasia. Am J Gastroenterol 1994; 89: 165-69.