CASE REPORT

Infantile Achalasia Cardia

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Background: Achalasia is extremely rare in infants. **Case characteristics**: We report three infants of age 9, 7 and 12 months, who presented with recurrent non-bilious vomiting, repeated chest infection and severe failure to thrive. Diagnosis of achalasia cardia was confirmed on contrast-swallow study. Heller's cardiomyotomy with fundoplication led to complete symptomatic relief, and weight-gain on follow-up. **Message**: Achalasia cardia is often misdiagnosed as gastroesophageal reflux disease which leads to significant delay in diagnosis and increased morbidity.

Keywords: *Infant, Heller's cardiomyotomy, Management, Vomiting*.

Achalasia cardia is rare in children, with a reported incidence of 0.11per lakh. Less than 5% of patients with symptomatic achalasia present below 15 years of age and of these less than 1 % are infants [1]. First case in an infant was reported by King in 1953 [2]. It is characterized by failure of lower esophageal sphincter (LES) to relax in response to swallowing. It is often misdiagnosed as gastroesophageal reflux disease (GERD), which may lead to significant delay in diagnosis.

CASE-REPORT

Case 1: A nine-month-old boy presented with recurrent non-bilious vomiting containing undigested feeds, repeated chest infections and failure to thrive (weight 4.5 kg, z score <–3 SD, height 70 cm). He had received treatment for GERD, but without any relief. Routine investigations revealed leucocytosis (62,900/mm^3) with lymphocytosis and positive procalcitonin. Cytomegalovirus (CMV) IgM and PCR were positive. He was started on intravenous Ganciclovir. Upper gastro-intestinal (UGI) endoscopy showed dilated esophagus. Barium swallow showed a dilated thoracic esophagus and narrowing at GE junction (***Fig.* 1**) suggestive of achalasia cardia. After optimization, he underwent Heller's cardiomyotomy with Dor's anterior fundoplication. At 6 months follow-up, baby had significant weight gain (weight 9 kg).

Case 2: This 18-month-old boy presented with complaints of recurrent lower respiratory tract infections and vomiting after feeds since the age of 7 months. He weighed 7.3 kg (z score <–2 SD, and height 81.2 cm). Routine investigations revealed leucocytosis (22,000/mm^3) and *X*-ray chest showed bilateral lung infiltrates. Contrast CT scan chest done outside revealed a dilated thoracic esophagus. Contrast swallow study showed characteristic 'bird beak' sign which confirmed the diagnosis of achalasia cardia (***Fig.* 2**) for which he underwent surgery. Six months post-operatively he had achieved weight of 10 kg.

Case 3: This 12-month-old presented with complaints of recurrent vomiting of uncurdled milk, since the age of 2 months. He weighed 6.5 kg (at–3 SD) and height was

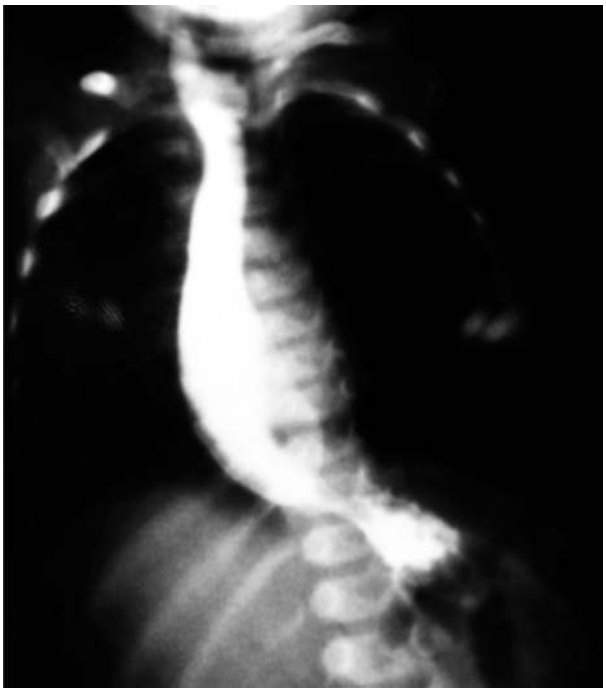

***FIG.* 1** *Upper GI contrast study in case 1, showing bird-beak sign.*

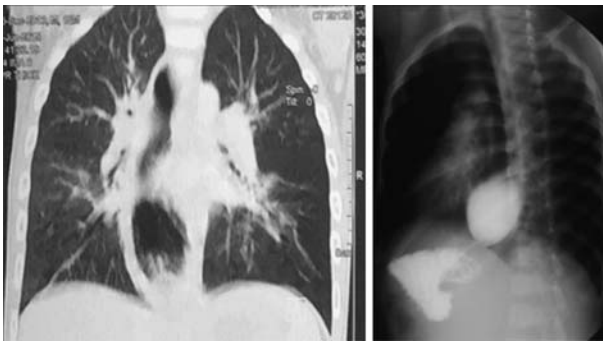

FIG. 2 *CECT chest showing dilated thoracic esophagus and upper GI contrast study showing megaesophagus in case 2.*

73.4 cm. Contrast swallow study showed classical 'bird beak' sign confirming the diagnosis of achalasia cardia. He underwent Heller's cardiomyotomy and had significant symptomatic relief post-operatively.

DISCUSSION

Achalasia cardia is a neuromuscular disorder characterized by degenerative changes of the myenteric plexus leading to selective loss of inhibitory nerve endings. This characteristically leads to failure of the coordinated muscle relaxation mechanism of LES after a peristaltic contraction of the esophageal body, stimulated by deglutition. Achalasia cardia can be classified as primary or secondary. Primary achalasia is a part of Allgrove and Alport syndrome, and is also seen in Down syndrome [3]. An association with some viral infections (Epstein Barr, herpes simplex and varicella zoster) has been described [3].

The most common characteristic feature of infantile achalasia is vomiting of uncurdled milk, which is also seen in regurgitation occurring commonly with faulty-feeding or over-feeding in infants and hence, it is usually missed initially. This kind of vomiting is also seen in infants with GERD. Frequently occurring complications of GERD such as aspiration pneumonia, esophagitis, midline chest pain, refusal to feed, failure to gain weight and anemia are also seen in achalasia cardia [4]. To rule out other esophageal disorders, UGI endoscopy followed by UGI contrast study is done. Manometry is the definitive investigation for diagnosis of achalasia cardia, but due to technical difficulties it is rarely done in infants [5].

Medical management of achalasia includes oral drugs like nitrates, calcium channel blockers and 5-phosphodiesterase inhibitors [5]. Pharmacological therapies provide temporary relief of symptoms and have not been recommended in infants [5]. Endoscopic injection of botulinum toxin has been used in adults but not preferred in infants [6]. Endoscopic balloon dilatation has been used in pediatric patients with achalasia but is not a preferred option due to associated complications such as perforation and short lasting effect [3]. Another non-surgical treatment modality described recently in adults and older children is per-oral endoscopic myotomy (POEM). It is a technically challenging procedure with 18% failure rate and high incidence of esophagitis (42%), esophageal perforation and GERD [6].

Heller's myotomy with anti-reflux procedure, is the gold standard for management of achalasia cardia. Symptomatic relief post-surgery is reported in 95% cases [6]. Although achalasia is rare in infancy yet it should be kept in mind if there is persistent regurgitation of feeds with failure to gain weight or repeated chest infections. It can be diagnosed easily by upper GI contrast study.

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REFERENCES

1. Chatterjee S, Gajbhiye V, De A, Nath S, Ghosh D, Das SK. Achalasia cardia in infants: Report of two cases. Journal of Indian Medical Association. 2012;44:1-5.
2. Sarangapani A, Vijayaraghavan S, Shanmuganathan S, Rajanikanth S. Achalasia cardia in infant. Sri Lanka J Infant Health. 2010;40:74-5.
3. Franklin AL, Petrosyn M, Kane TD. Infanthood achalasia: a comprehensive review of disease, diagnosis and therapeutic management. World J Gastrointest Endosc. 2014;6:105-11.
4. Rai GS, Sarawagi R, Sharma S, Apte A. An infant with nasal regurgitation since birth and failure to thrive. J Clin Diagn Res. 2015;9:3.
5. Dughera L, Chiaverina M, Cacciotella L, Fabio C. Management of achalasia. Clin Exp Gastroenterol. 2011;4:33-41.
6. Patti MG, Fischella PM. Controversies in management of Achalasia. J Gastrointest Surg. 2014;18:1705-9.