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Case Report

Achalasia cardia in an infant - were we too late?

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ABSTRACT

Achalasia cardia is a motility disorder where there is aperistalsis of the body of the esophagus and failure of lower esophageal sphincter relaxation. This manifests as a functional gastroesophageal junction obstruction. This disorder is rare and has a prevalence of 10 in 100,000 population. It has a peak incidence between 30-60 years of age. The incidence in the pediatric population is very low and is even rarer in infants. Here we present a case of achalasia cardia in a 9-month-old male infant weighing 3.1 kilograms. The child had complaints of regurgitation of milk, non-projectile vomiting, fever and cough for the past 3 months along with failure to thrive. He was treated for bronchopneumonia initially and then evaluated with a barium swallow. The barium swallow revealed a hold-up of contrast in the distal esophagus along with a typical bird-beak appearance. Definitive surgical intervention was being planned. However, the child aspirated and expired. Did we delay the procedure? Were we too late? These questions continue to make us self-introspect on the management of infantile achalasia cardia. The paucity of available publications and data is a major roadblock in management.

Keywords: Achalasia cardia, Infant achalasia, Myotomy

INTRODUCTION

Achalasia cardia is a well-documented motility condition that affects the lower esophagus and is marked by a characteristic lack of lower esophageal sphincter (LES) relaxation and aperistalsis in the body of the esophagus. Among the motility disorders of the esophagus, achalasia cardia is the best understood. The incidence and prevalence vary amongst populations but it is not a common disease. Even in areas of high prevalence, incidence rates have been documented to be around 1.63 per 10000 people.[1] There is no strong sex predilection although certain studies have demonstrated slightly higher incidence in females. The incidence peaks at the age of 30-60 years. Achalasia cardia is no doubt a rare disease but even more rare when it is present in children and infants.

The basic pathophysiology is poorly understood but it is believed that an inflammatory neurodegenerative insult with a possible viral component is likely. Autoimmune disorders such as SLE, Sjogren's disease are associated with an increased risk of Achalasia. Chagas' disease affecting the esophagus also demonstrated features similar to achalasia.

The presence of a classical barium study showing a bird's beak appearance is usually diagnostic. Like in adults, the gold standard of diagnosis remains esophageal manometry. However, the lack of established protocols limits its use. The need for sedation in younger children and infants also obscures the findings.[2]

Children often present with dysphasia, vomiting and weight loss. Younger children come to us with failure to thrive, nocturnal cough, recurrent pneumonia, and feeding difficulties.[3] Like in adults, management of achalasia includes calcium channel blockers, botulinum toxin injection, per-oral endoscopic myotomy and surgery. The

limited literature we have currently suggests that surgical management using a laparoscopic Heller's myotomy remains the most effective mode of treatment.

CASE REPORT

A 9-month-old male child weighing 3.1 kilograms (weight less than 3rd percentile for age) reported to the pediatric emergency with complaints of regurgitation of food, recurrent non-projectile vomiting, fever and cough. The child had regurgitation after every feed for the past 3 months. The child was initially treated at a local hospital as a case of suspected gastroesophageal reflux disease and had been advised feeding in an upright position but symptoms still persisted. There had been previous two hospitalizations for bronchopneumonia. The child was finally referred to our tertiary care centre after recurrent episodes of pneumonia and failure to thrive. The child was poorly nourished with a significant loss of subcutaneous fat and muscle.

The infant is a first-born male child born out of non-consanguineous marriage. Delivery was via caesarean section at term. The child was very low birth weight, weighing just 1.25 kilograms at birth and was immediately shifted to the new-born intensive care unit for distress and low birth weight. He was discharged after 4 weeks of hospital neonatal care and exclusively breastfed for 1 month after which there was lactation failure in the mother (possibly due to low birth weight). The infant displayed a delay in developmental milestones. He is not able to sit without support and has a persisting bidextrous grasp.

Management and outcomes

Blood work up and serology were unremarkable except for mild anemia. The blood picture was characteristic of nutritional anemia. X ray chest on arrival showed a dilated esophagus and no fundal gas shadow. There were bilateral patchy opacities in the lung fields.

The child was treated with intravenous antibiotics and stabilized. Nasogastric feeds were initiated. The child improved over a span of 4 days. A barium swallow was then performed as we suspected an esophageal motility disorder. Our suspicion was prompted by the presence of a dilated esophagus along with the absent fundal gas shadow. The barium swallow revealed a typical bird beak appearance with a smooth narrowing of distal esophagus (Figure 1).

The upper GI endoscopy study was performed soon. It revealed partially digested food particles in the esophagus. We considered esophageal manometry but the need for sedation, lack of established protocols and lack of available expertise in pediatric manometry led us to reconsider it. We were in discussion with the pediatric surgery team to plan for surgery when the child suffered an episode of aspiration and expired soon after.

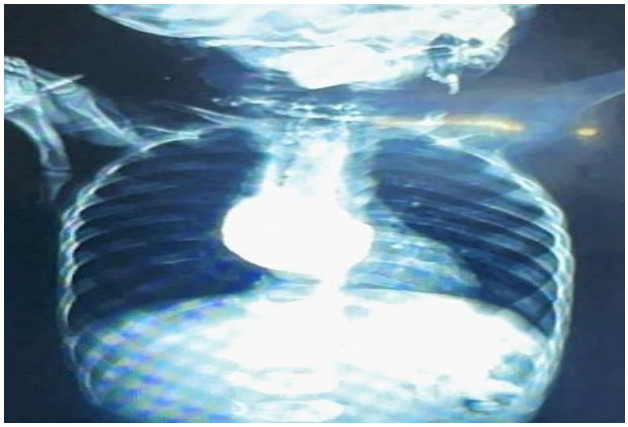

Figure 1: Barium swallow of child with bird beak appearance and distal hold-up of contrast.

DISCUSSION

The outcome in this case was very distressing. The amount of published literature about achalasia cardia in the pediatric age group, even more so in infants, is very less. Despite being the most common esophageal motility disorder, it is not an entity reported often in infants.[4] The incidence in the pediatric age group is less than 0.11/100000 children. The characteristic feature of achalasia cardia is the failure or uncoordinated relaxation of the LES along with relaxation of the esophageal body. It is understood that the loss of inhibitory ganglions in the myenteric plexus of the esophagus is the basic pathology. The loss of inhibition initially leads to over-excitation of the cholinergic neurons. This results in high amplitude non-peristaltic contractions (vigorous achalasia). Progressive loss of cholinergic neurons slowly results in low amplitude simultaneous contractions in the esophageal body (classic achalasia).[4] The exact cause of this disorder cannot be pinpointed however a plethora of genetic, environmental, autoimmune and infective factors have been implicated. Chagas' disease often has features that mimic achalasia. Achalasia is known to be a part of the spectrum of Allgrove syndrome, Alport syndrome and Down syndrome.[5] A genetic basis for achalasia cardia has been attributed to the fact that is the noted incidence in monozygotic twins, siblings and 1st degree relatives.[4] The triple A syndrome or Allgrove syndrome as it is popularly known is caused by mutations in the AAAS gene. This gene encodes a protein known as alacrima achalasia adrenal insufficiency neurologic disorder (ALADIN).[6] This novel protein is well researched and documented. However, a variety of mutations such as nitric oxide synthase polymorphisms, VIPR1 gene polymorphisms, interleukin-23 receptor polymorphisms, protein tyrosine phosphatase non-receptor 22 gene polymorphisms are known to be associated with achalasia cardia.[4] Infective causes such as varicella zoster and Epstein-Barr virus are also associated with achalasia cardia.

In the Indian scenario, vomiting of uncurdled milk is often the most common presentation.[5] Children often present

with pneumonia secondary to aspiration of feeds. Most children come to us with poor growth parameters. Vomiting of uncurdled milk is also seen in regurgitation due to faulty feeding and overfeeding. This is one of the suggested reasons for misdiagnosis. Another important differential diagnosis is gastroesophageal reflux disease (GERD). Vomiting of uncurdled milk, features of aspiration pneumonia, failure to thrive are also seen in GERD.

Medical management usually comprises nitrates, calcium channel blockers and 5-phosphodiesterase inhibitors taken orally. Their role is limited though in infants and children.[5,7] Botulinum toxin is often used in adults as a first line therapy but there has been no evidence to support the use in infants.[8] Endoscopic management using per-oral endoscopic myotomy (POEM) is known to have good results in older children and adults but no such evidence exists in infants. Surgery using Heller's cardiomyotomy is the gold standard of care. It can be performed either laparoscopically or via open surgery. There have been multiple case series from India which have reported successful management of achalasia cardia by surgery. Chatterjee et al report 2 cases who were surgically treated with Heller's cardiomyotomy along with a partial Toupet fundoplication.[9] Both the infants recovered well and gained weight. Banerjee et al report a series of 3 cases successfully treated with Heller's cardiomyotomy.[5] The surgery involves a longitudinal incision in the esophageal musculature approximately 5 cm above the esophagogastric junction, extending 2-3 cm onto the cardia of the stomach. The laparoscopic approach is considered superior to open surgery due to reduced pain and faster recovery.[3]

CONCLUSION

Our patient should have undergone surgery early in the course of his disease. A delayed diagnosis in the peripheral set-up meant that the infant had undergone recurrent episodes of bronchopneumonia. The child also had failed to thrive. The rarity of the disease means that a lot of children get misdiagnosed as GERD. Early referral to tertiary centres will help save lives. Greater awareness about achalasia cardia among treating pediatricians might lead to earlier referrals and appropriate treatment.

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