Niger J Paediatr 2023; 50 (3):151 – 155

CASE REPORT

Osarogiagbon O. Wilson
Efobi A. Chizoba
Eyo-Ita U. Emmanuel
Benjamin Nandom

Achalasia cardia: an uncommon cause of aspiration syndrome in infancy

DOI:http://dx.doi.org/10.4314/njp.v50i3.4

Accepted: 21st September 2023

Osarogiagbon O. Wilson (✉)
Efobi A. Chizoba,
Eyo-Ita U. Emmanuel,
Benjamin Nandom
Department of Child Health,
University of Benin Teaching Hospital, Benin city.

Abstract: Achalasia is a primary oesophageal motility disorder characterized by the absence of Oesophageal peristalsis and impaired lower oesophageal sphincter (LES) relaxation in response to swallowing leading to functional obstruction at the gastroesophageal junction. It is unusual in childhood and extremely rare in infants. We report a case of Achalasia cardia in a twomonth bold infant treated with anterior oesophago-cardio-gastromyotomy (Modified Heller's procedure).

Introduction

Achalasia is a primary oesophageal motility disorder characterized by absence of oesophageal peristalsis and impaired lower esophageal sphincter (LES) relaxation in response to swallowing leading to functional obstruction at the gastroesophageal junction.[1] It is a rare oesophageal neurodegenerative disorder in the paediatric population. The first case of the condition was reported by Sir Thomas Willis in1674. He described oesophageal dilation with a whale bone in a patient who had dysphagia and a dilated oesophagus. The term achalasia itself was coined by Hurst in the year 1927. Achalasia is extremely rare in infants and scarcely occurs in children under five years of age. The overall annual incidence in children is 0.11/100,000. Less than 5% of patients with symptomaticachalasia present before their 15th birthday.[2] However, our patient was two months old at the time of diagnosis.

Case report

A twomonth old male infant who presented with a three-week history of poor weight gain and cough, fever and difficulty with breathing of two weeks and a convulsion which began five days before presentation. Following the onset of coughing, the baby was taken to a General Hospital where he was managed on outpatient basis with a 'cough syrup' for three days. The fever was described as his body being very hot; this persisted through most of the day with transient relief whenever he was given oral paracetamol. With persistence of the cough and onset of difficulty with breathing, he was then taken to a private hospital where the clinical diagnosis of pulmonary tuberculosis was made; he was admitted for eleven days and treated with intravenous ceftriaxone, oral Quinineand oral anti- tuberculosis medications (syrup Rifampicin and Isoniazid). He was discharged home on the anti-tuberculosis medications. His mother however discontinued the treatment four days after because she had not noticed any difference in the child's condition.

He remained at home for one week during which symptoms worsened. He was then referred to UBTH from a private hospital with worsening of his symptoms.

At presentation in UBTH he was critically ill-looking, in obvious respiratory distress, febrile (38^{o}C), pale and, underweight (76% of expected).

He was tachypnoeic and dyspnoeic with a respiratory rate of 80 cycles/min and marked flaring of the alae nasi, intercostal and sub-costal retractions. Vesicular breath sounds were heard with wide spread crepitations and rhonchi worse in the right upper lung zone. SpO2 was 89% in room air. His heart rate 160bpm.

He was unconscious (Blantyre Coma Scale score = 3/5: Eye Opening =1; Verbal response = 1; Motor activity = 1), had no facial asymmetry. His pupils were equal, central and, briskly reactive to light bilaterally. Tone was reduced globally, deep tendon reflexes were reduced globally and his neck was supple.

His abdomen was full, moved with respiration and soft. He had soft, tender hepatomegaly (liver span of 12cm). Other aspects of the systemic examination were unremarkable.

His chest radiograph showed bilateral peri-hilar infiltrates and a cardiothoracic ratio of 60%.

The working diagnosis was sepsis with suspected Meningitis, bronchopneumonia with congestive Cardiac failure and disseminated tuberculosis (Tb meningitis, Pulmonary Tb). He was started on intravenous cefotaxime, genticin and intravenous fluids at 75% maintenance. While on admission, the fever resolved but the coughing and difficulty with breathing persisted.

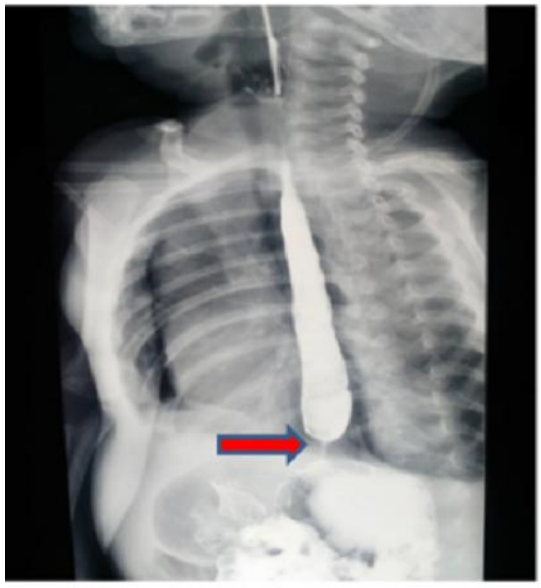
The respiratory symptoms often worsened when attempts were made to change from NGT feeding to oral feeds. He suffered two apnoeic episodes during milk feeds with subsequent worsening of respiratory signs and chest radiographic findings of patchy opacities especially in the right hemithorax.

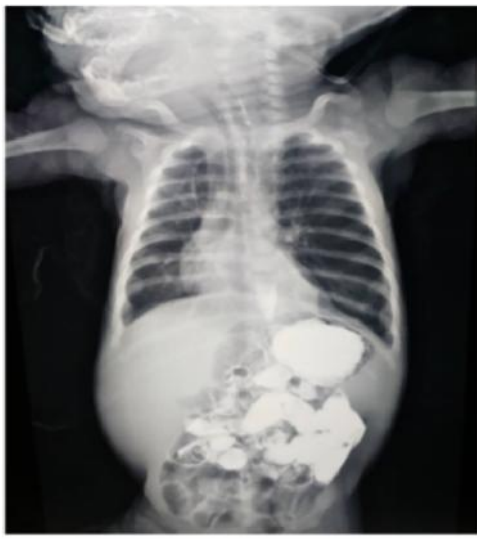
The diagnosis of an aspiration syndrome was considered; possibly gastro-esophagealreflux disease, H-type

tracheo-oesophageal fistula and achalasia of the cardia.
Complete haemogram revealed anaemia (packed cell volume = 26.6%) and leucocytosis, with differential neutrophilia. Serum electrolytes, urea and creatinine showed hyponatraemia and hypokalaemia while the erythrocyte sedimentation rate was normal, mantoux test was non-reactive. Cerebro-spinal fluid (CSF) analysis was normal and stool and CSF for Xpertmtb/Rif detected no MTB. The chest radiograph showed bilateral peri-hilar infiltrates and barium meal revealed persistent luminal narrowing involving the distal part of the oesophagus with mild dilatation of its proximal part. No evidence of oesophageal masses, fistula or mucosal ulceration was seen. There was no evidence of laryngeal penetration or aspiration. These features were consistent with aganglionosis of the distal oesophagus.
After correction of anaemia, dyselectrolytaemia and other parameters, the child had modified Heller's procedure.

Intra-operative findings were slight constriction of the cardia, otherwise normal sized oesophagus. He was commenced on IV cefuroxime and inhaled fluticasone. Following return of bowel sounds, he was started on oral fluid (expressed breast milk) and oral metoclopramide.
Barium swallow done post intervention showed normal caliber middle and lower esophagus with no demonstrable narrowing of the lower oesophagus lumen.
The patient was discharged with complete resolution of symptoms. He is currently being followed up at the paediatric respiratory and cardio-thoracic surgical clinics. His weight is now appropriate for age. There has been no recurrence of the symptoms.

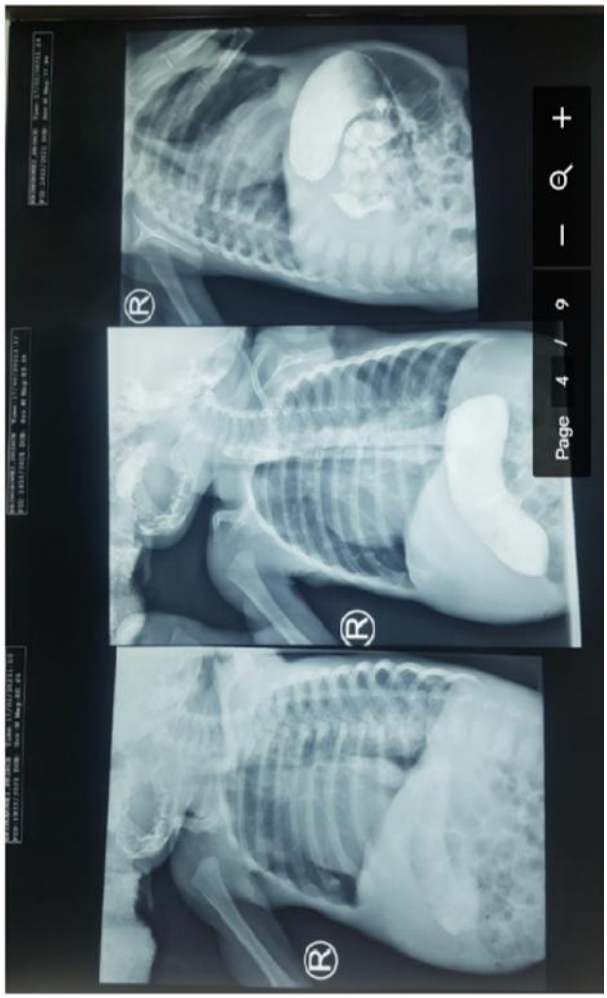
Barium swallow (Before surgical intervention)





The histology report of tissue specimens from the lower oesophageal wall revealed features in keeping with Achalasia viz; hypertrophied muscularis propria and hypertrophied nerve bundles of the Auerbach plexus with absent ganglion cells. There were also areas of fibrosis within the muscularis propria and serosa with inflammatory cells, predominantly lymphocytes.

Barium swallow (After surgical intervention)



Discussion

Achalasia cardia is extremely rare in infants. Affecting 0.5 to 1.0 in 100,000 persons per year, its prevalence has been estimated to be 8 per 100,000. The incidence of achalasia cardia in Asian and African populations is lower than in the British populations (0.3 per 100,000).[3] Less than5% of patients with symptomatic achalasia present before the age of 15 years and of these, less than 1% are infants. The mean age at diagnosis in children is 8.8 years, with a mean duration of symptoms before diagnosis of 23 months. Our patient was an infant and had started havingsymptoms a few weeks before presentation. Achalasia is often misdiagnosed as gastro-oesophageal reflux disease (GORD) causing significant delays in making appropriate diagnosis and institution of needful intervention. A similar trend was noted in our patient whose diagnosis was delayed for weeks as other differentials were considered and ruled out. Though our patientwas male, no sex predilection has been reported in literature.[4]
GORD was considered in our patient as it usually manifests within the first few months of life, peaks at about four months with most cases resolving before one year and almost all by two years.
The aetio-pathogenesis of Achalasia is poorly understood. Advanced theories include:

1. *Degeneration of the Auerbachs plexus by Autoimmune inflammation:*

This is supported by the presence of circulating myenteric antibodies and inflammatory T-cellinfiltrates in myenteric plexus of patients with achalasia. Also, patients with achalasia are 3.6times more likely to have other autoimmune diseases. Serum from achalasia patients (but not from those with GORD) can induce phenotypic and functional changes in myenteric neurons and reproduce the characteristics of the disease.[5]

2. Infectious (Chagas disease or viral infections) diseases

American trypanosomiasis or Chagas disease is a vector-borne disease caused by the protozoan *Trypanosoma cruzi.* Its natural vectors are blood sucking insects of the family Reduviidae. It manifests acutely with non-specific febrile illness and in the chronic state is associated with cardiomyopathy and severe gastrointestinal abnormalities. Two main mechanisms are likely involved-direct tissue destruction by low-level parasite persistence mediated by lymphocyte infiltration and fibrosis and molecular mimicry of host antigens by the parasite, resulting in autoantibodies that produce an inflammatory reaction associated with direct tissue damage and direct stimulation of adrenergic and muscarinic cholinergic receptors associated with dysautonomia and increased risk of arrhythmia. In patients with gastrointestinal tract involvement, myenteric plexus destruction leads to pathologic sign of organ dilation [5]
May also be familial and a part of the All groove syndrome (achalasia, alacrima and corticotrophin insensitivity).[6]

Achalasia is a primary oesophageal motility disorder with degeneration of the inhibitory myenteric plexus innervating the lower oesophageal sphincter and esophageal body leading to an imbalance in inhibitory and excitatory neurons resulting in failure of the lower oesophageal sphincter to relax with swallowing, absence of peristalsis of the oesophageal body and increased resting pressure of the sphincter. The most common characteristic feature of Infantile Achalasia is vomiting of uncurdled milk which is also seen in regurgitation resulting from faulty feeding or over feeding in infants thus it is usually missed initially. Frequently occurring complications such as aspiration pneumonia, oesophagitis, midline chest pain, refusal to feed, failure to gain weight and anaemia are also seenin achalasia cardia. Our patient had recurrent episodes of aspiration pneumonitis but was probably too young to show clear features of oesophagitis and it is possible that the movements described as convulsions were actually opisthotonic movements and jerks that resulted from oesophagitis. In severe cases patients can present with features suggestive of obstructive apnoea or with stridor or lower airway disease. The index patient had wheezing and features of lower airway disease including; marked dyspnoea crepitations, and rhonchi.

Following massive aspiration, patients may develop cyanosis and pulmonary oedema which may progress to severe respiratory distress syndrome. These were also seen towards the third week on admission after the patient's meals had been increased to 100mls/kg/day and after discontinuation of NG tube feeding.
The chest radiograph in Achalasia typically shows an air-fluid level within a dilated oesophagus. Barium fluoroscopy reveals smooth tapering of the lower oesophagus leading to the closed lower oesophageal sphincter resembling a bird's beak. Loss of primary peristalsis in the distal oesophagus with retained food and poor emptying of proximal contents create this abnormal shape of the oesophagus.

The ostensibly unexplained poor response to therapy in our patient tilted our diagnostic probe in search of a chronic aspiration syndrome. He had a barium swallow that revealed distal oesophageal tapering with mild dilatation proximal to the narrowing. Also noted were hyperinflated left lung with a curvilinear opacity in the right upper lobe.
Manometry is the most sensitive diagnostic test; it reveals the defining features of aperistalsis in the distal oesophageal body and incomplete or absent lower oesophageal sphincter relaxation often accompanied by high pressure lower oesophageal sphincter and low amplitude oesophageal body contractions. This was not done for our patient as the necessary equipment is not available at our facility.

The two most effective treatment options are pneumatic dilation and laparoscopic or surgical (Heller) myotomy.
Pneumatic dilation is the initial treatment of choice. Here a balloon catheter is inserted into the lower oesophageal sphincter and inflated to force it to dilate.[7,8]
Laparoscopic or surgical (Heller) myotomy plus fundoplication (Modified Heller's procedure) is an anti-reflux procedure. This prevents the development of GORD as a post-surgical complication which can occur after the sphincter has been rendered less competent.[9--12]

Our patient had a modified Heller's procedure done with anterior fundoplication. Intra-operative finding was a constriction of the distal oesophagus.
It occurs due to a reduced tone of the lower oesophageal sphincter, abnormal frequency of sphincter relaxation and hiatal hernia. Infant reflux manifests as regurgitations, oesophagitis, (irritability, cough, arching, choking, gagging and feeding aversion) as well as failure to thrive. These symptoms may also have been what were interpreted as convulsions earlier in our patient. In severe cases it can manifest as obstructive apnoea or as stridor or lower airway disease.
Diagnosis is by oesophageal pH monitoring (pH < 4); endoscopy to visualize the sphincter and associated ulcers; empirical anti-reflux therapy using proton pump inhibitors. Detection of pepsin in tracheal fluid is a marker of reflux-associated aspiration of gastric contents.
GORD was considered as a probable causes of aspiration in our patient as it usually manifests within the first few months of life, peaks at about four months and our patient was a twomonth old male who had cough, difficulty with breathing with pneumonic changes on chest radiography.

Tracheoesophageal fistula (H-type) and oesophageal atresia were also considerations in this patient. They are the most common congenital anomaly of the oesophagus with a prevalence of 1.7 per 10,000 live births. About 90% of oesophageal atresia cases are associated with tracheoesophageal fistula. The exact cause is not known but associated features include advanced maternal age, European ethnicity, obesity, low socioeconomic status and tobacco smoking, all of which were absent in the index patient.
They typically present with frothy secretions from the mouth and nose after birth as well as episodes of coughing, cyanosis and respiratory distress. Feeding worsens symptoms, due toregurgitation of feeds or direct aspiration through the fistula causing recurrent pneumonia. The index patient had recurrent episodes of aspiration pneumonia but was not found to have frothy mouth or nasal secretions.
In the H-type tracheo-oesophageal fistula, an oesophagogram with contrast injected under pressure can demonstrate the defect. Surgical ligation of the fistula and primary end to end anastomosis of the oesophagus via a right–sided thoracotomy constitute the current standard of surgical approach for treatment.

Conclusion

Achalasia cardia is rare in infancy. A high index of suspicion is required for diagnosis as its presentation is similar to other aspiration syndrome, principal among which is Gastro oesophageal reflux disease (GORD) in infancy. Early diagnosis and prompt intervention is necessary for a successful outcome.

References

1. Patti MG, Fisichella PM, Perretta S, Galvani C,Gorodner MV, Robinson T, et al.. Impact of minimally invasive surgery on thetreatment of esophageal achalasia: a decade of change. *J Am Coll Surg. 2003; 196:698 –703. http:// dx.doi.org/10.1016/S1072-7515 (02)01837-9.*
2. Hamza AF, Awad HA, Hussein O. Cardiac Achalasia in Children. Dilatation orSurgery? *Eur J Pediatr Surg. 1999; 9:299–302. http:// dx.doi.org/10.1055/s-2008-1072268.*
3. Reynolds JC, Parkman HP. Achalasia Gastroenterol Clin North Am. 1989; 18:223–55.
4. Marlais M, Fishman JR, Fell JME, Haddad MJ, Rawat DJ. UK incidence of achalasia: an 11-year nationalepidemiological study. *Arch Dis Child. 2011; 96:192–194. http:// dx.doi.org/10.1136/ adc.2009.171975.*
5. Kleigman R, Stanton B, St Geme JW, Schor NF, Behram RE, Nelson WE.Textbook of paediatrics;21st edition(2019)
6. Boeckxstaens GE, Annese V, Bruley des Varannes S, Moonen A, Belmans A, Costantini M, et al. The European achalasia trial: arandomized multi -centre trial comparing endoscopic pneumodilation and laparoscopic Hellermyotomy as primary treatment of idiopathicachalasia. *Gastroenterology. 2010; 138* (Suppl1):S53. http://doi.org/ c2zh2v.
7. Cloud DT, White RF, Linkner LM, Taylor LC. Surgical treatment of esophageal achalasia in Children. *J Pediatr Surg. 1966; 1:137–44.*
8. Murray G, Battaglini JW, Keagy BA, Starek PJ, Wilcox BR. Selective application of fundoplication in Achalasia. *Ann Thorac Surg. 1984; 37:185–8. http:// dx.doi.org/10.1016/S0003-4975(10)60321-9.*
9. Sonnenberg A, Massey BT, McCarty DJ,Jacobsen SJ l. Epidemiology of hospitalization for achalasia in the United States. *Dig Dis Sci. 1993; 38:233–44. http:// dx.doi.org/10.1007/ BF01307540.*

10. Tachovsky TJ, Lynn HB, Ellis FH., Jr The surgical approach to esophageal achalasia inchildren. *J Pediatr Surg. 1968; 3:225–31. http://doi.org/ cq4x4r.*

11. Moazam F, Rodgers BM. Infantile achalasia. Brief clinical report. *J Thorac Cardiovasc-Surg. 1976; 72:809–12.*

12. Hunter JG, Trus TL, Branum GD, Waring JP. Laparoscopic Heller myotomy andFundoplication for achalasia. *Ann Surg. 1997; 225:655–64. discussion 664–65. http://doi.org/db33z.*