

Oesophageal achalasia in an infant- A rare case report

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Abstract: Achalasia cardia is a neuromuscular disorder of the oesophagus, characterized by abnormal oesophageal motility and failure of lower esophageal sphincter relaxation. It usually manifests in the fourth and fifth decades of life. The mean age in children is 8.8 years with a mean duration of symptoms before diagnosis of 23 months. It is unusual in childhood and is extremely rare under the age of one year. We report a 9-month old female child with oesophageal cardia treated with oesophagomyotomy with anti-reflux procedure via the abdominal route.

Keywords: Achalasia cardia, dysphagia, Gastro esophageal reflux disease (GERD), infant, oesophagomyotomy.

INTRODUCTION

Achalasia is characterized by failure of coordinated relaxation of lower oesophageal sphincter (LES) combined with ineffective peristalsis in body of the oesophagus, resulting in functional obstruction of the oesophagus [1-5]. Esophageal achalasia of unknown etiology is a rare disease, affecting 1:100,000 of the general population and just 4 to 5% of these cases are described in children, [6] and less than 1% in infants. The symptom spectrum is wide, varying from regurgitation to recurrent aspiration pneumonia. Older children describe symptoms of progressive dysphagia. For these reasons the disease is often confused with gastro-esophageal reflux disease (GERD), delaying correct diagnosis [7-8]. In response to the rarity of the case in infancy, we present a case of oesophageal cardia in an infant.

CASE REPORT

A 9-month old first born female child of non-consanguineous parents presented to us with regurgitation of feeds and poor weight gain since 6 months of age. She used to vomit unaltered feeds immediately after ingestion. She was hospitalized twice in previous two months for pneumonia. She was treated as GERD with pneumonia with intravenous antibiotics and anti GERD treatment. She was discharged after seven days of treatment. There was no significant birth/neonatal/family history related to her illness. On examination, the infant was malnourished and anemic with a weight of 7.6kg and length of 68cms. Examination of other systems was unremarkable. Routine blood investigations were within normal limits but with hemoglobin of 9gm/dl. Barium swallow showed tapering of the oesophagus close to the cardia

(Fig. 1) and dilated oesophagus (Fig. 2). Motility study could not be done because of non-availability of this study in our hospital. A diagnosis of achalasia cardia was based on clinical grounds and by barium swallows study. The infant underwent Heller's oesophagocardiomyotomy with fundoplication.

The baby was started on oral feeds on 3rd post operative day. Post operative period was uneventful. She was discharged after seven days. Her follow up examination revealed dramatic relief of symptoms and satisfactory weight gain.



Fig. 1: Oesophagogram showing tapering of the oesophagus close to the cardia



Fig. 2: Oesophagogram showing severe dilatation of the oesophagus

DISCUSSION

Thomas Willis first described achalasia in 1674. He successfully treated a patient by dilating the lower esophageal sphincter (LES) with cork-tipped whalebone. Esophageal achalasia is a failure of the coordinated muscle relaxation mechanism of the LES after a peristaltic contraction of the esophageal body, stimulated by a deglutition [7]. The etiology of the disease is unknown and it is rare during childhood, and it affects more male children (6:1) than female, [7-10] while in the case described above the patient was female.

The following theories can explain the onset of the disease: (1) a primary neurogenic abnormality with a failure of the inhibitory nerves and progressive degeneration of ganglion cells; (2) an acquired deficiency of the myenteric plexus ganglion cells, secondary to GERD, Chagas disease or viral process [6, 9] Degenerative, autoimmune (antibody to Auerbach's plexus), and infectious factor are possible causes [10]. Achalasia cardia can occur as a part of Allgrove syndrome, where it is associated with alacrimia and adrenocorticotrophin hormone insensitivity [11, 12]. There is a selective loss of post-ganglionic inhibitory neurons that normally lead to sphincter relaxation, leaving post ganglionic neurons unopposed. This imbalance produces high basal LES pressures and insufficient LES relaxation. The loss of oesophageal peristalsis can be a secondary phenomenon. Typically, adults and older children have progressive dysphagia to solids and liquids, chest pain weight loss [11-18] and heart burn [19]. But, symptoms in infants and toddler are choking, cough, recurrent pneumonia and failure to thrive [12-17]. Retained oesophageal food can produce oesophagitis. [10] Vomiting of uncurdled milk is characteristic of achalasia [20]. As the clinical manifestations of achalasia cardia mimic other causes of esophageal obstruction, it challenges the clinical acumen of the attending physician [21].

The classical oesophagographic or barium fluoroscopy finding for diagnosis of achalasia cardia is bird's beak appearance [22]. Upper gastrointestinal endoscopy is helpful in the diagnosis of achalasia cardia as well ruling out severe oesophagitis and malignancy. Scintillographic studies and oesophageal tests are useful to diagnose or to rule out GERD [7-9]. Oesophageal manometry is optional to confirm the presence of achalasia and reveals incomplete relaxation of a high pressure LES during swallowing, often accompanied by non propulsive simultaneous contractions in the distal oesophagus. We were unable to do this examination, owing to the lack of this facility. If biopsy of distal oesophagus is done, lack of myenteric plexus innervation can be demonstrated [11].

The two most effective treatment options are pneumatic dilatation and surgical (Heller's cardiomyotomy. Surgeon often supplement a myotomy with an anti reflux procedure to prevent the GERD that otherwise often ensues when the sphincter is rendered less competent [11]. Recurrence of symptoms within 6 months and oesophageal perforation are the main concerns of the procedure following pneumatic dilatation of the oesophagus. [23, 24]

The classical surgical approach is myotomy for the oesophagus and cardia. This extends from the LES to 2 or 3 cm above the cardia, generally associated with anti reflux procedures, and it is done with a view to the fact that cardiomyotomy alone favors GERD [7, 21].

In recent years, laparoscopy has made surgical treatment of esophageal achalasia less invasive. The approach can be either abdominal or thoracic. According to reports, [24, 25] the so-called minimally invasive surgery (laparoscopic) does not just represent the most effective treatment for the disease, it also reduces the time of hospitalization (an average of 2 to 3 days) and hospital costs, allowing the child to recover more quickly. Pharmacotherapy includes the use of smooth muscle relaxants like isosorbide dinitrate or calcium channel blockers like nifedipine or local injection of botulinum toxin which has been tried with some success in adults [6, 9] It counterbalances the selective loss of inhibitory neurotransmitters by inhibiting the release of acetylcholine from nerve terminals [11].

In the case described here we opted for Heller's cardiomyotomy with Nissen fundoplication achieving good post operative progress and early hospital discharge and with no sign of relapse to date.

CONCLUSION

Oesophageal achalasia is a rare disease in children and its origin is generally indeterminable. It presents a diagnostic challenge in infancy. The oesophagogram can precisely confirm the diagnosis.

Heller's cardiomyotomy is the treatment modality of choice and which resulted in good clinical progress. Infants may predominantly have respiratory symptoms which may overshadow the vomiting, as in our case. Early diagnosis and surgical oesophagomyotomy leads to a successful outcome.

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