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CASE REPORT

Cardiac Achalasia in Infancy.

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ABSTRACT

Background: Achalasia is one of the rarest illnesses of the esophagus. Adults are more prone to cardiac achalasia. Occasionally it may occur in adolescence and late childhood, but it is very rare in infancy. The diagnosis of early-onset cardiac achalasia depends mainly on the symptoms that the patient complains of. The main symptom is retrosternal discomfort or dysphagia. These symptoms can't be informed by a nonverbal infant.

Case presentation: We have described an 11-month-old boy with cardiac achalasia who presented with recurrent vomiting, recurrent chest infection and failure to thrive. Our case was treated with Heller's esophagocardiomyotomy and an anti-reflux procedure.

Conclusions: Cardiac achalasia is regarded as a rare disease in infants. Misdiagnosis as GERD is common. We should suspect this disease especially if the infant has recurrent RTI, persistent vomiting, and failure to thrive.

Key words: cardiac achalasia; vomiting; infant

INTRODUCTION

Cardiac achalasia is one of the rarest illnesses of the esophagus [1]. Adults are more prone to cardiac achalasia. Occasionally it may occur in adolescence and late childhood, but it is very rare in infancy [2]. The disease is even more infrequent in children less than 5 years of age. The incidence of achalasia in childhood is 0.11/100000 children annually [3,4] and less than 5% of patients having symptoms their age are less than 15 years [5].

Cardiac achalasia is a primary motility disorder of the esophagus which is characterized by the absence of effective peristalsis in response to swallowing and a failure of relaxation of the lower esophageal sphincter (LES) [6]. The actual cause of cardiac achalasia is unknown. The defect in peristalsis may occur due to degeneration of the myenteric nerve plexus of the esophagus or ganglion cells may be not present [7,8].

The diagnosis of early-onset cardiac achalasia depends mainly on symptoms from which the patient complains. The main symptom is retrosternal discomfort or dysphagia [9]. These symptoms can't be expressed by a nonverbal infant. Repeatedly the parents of the infant describe the expulsion of milk from the mouth as vomiting instead of regurgitation making the diagnosis so difficult [10]. Failure to thrive or gradual loss of weight and recurrent respiratory tract infection with cough during sleep are nonspecific manifestations for diagnosis of cardiac achalasia. No specific signs were present during Physical examination [10]. Barium swallow remains one of the most important tools for the diagnosis of achalasia [2].

CASE REPORT

Written informed consent was obtained from participant parents, the study was approved by the research ethical committee of the Faculty of Medicine, Zagazig University. The study was done according to The Code of Ethics of the World Medical Association (Declaration of Helsinki) for studies involving humans.

An 11-month – old infant presented with vomiting dated since birth. Vomiting was non-bilious, non-projectile occurred after most of the feeds, and the baby was malnourished (body weight was 6.5 kg, below the third centile). The baby suffered from respiratory tract infection 5 times since birth. History of delayed motor and mental milestones. Physical examination showed moderate dehydration

and pallor but there was no jaundice or cyanosis. Abdominal examination was normal, with no visible peristalsis, abdominal distention, or organomegaly

Our provisional diagnosis was Gastroesophageal reflux disease (GERD). The investigations done included complete blood count (CBC), serum electrolytes, blood urea, serum creatinine, blood sugar, and liver functions were within normal limits. Abdominal ultrasonography was done to exclude hypertrophic pyloric stenosis and other congenital anomalies of the duodenum and stomach. A Series of X-rays with contrast were done to GIT (barium swallow, barium meal, and barium follow-through). Cardiac achalasia was diagnosed from the mega-esophagus and smooth tapering appearance of the lower end of esophagus like bird's beak appearance with retained contrast.The patient was referred to the surgical unit and prepared for esophagiocardiomyotomy operation. Following surgery oral nutrition was permitted on the 2nd postoperative day, and there was no problem with vomiting or regurgitation after feeding. Follow up of the baby, the boy was growing well

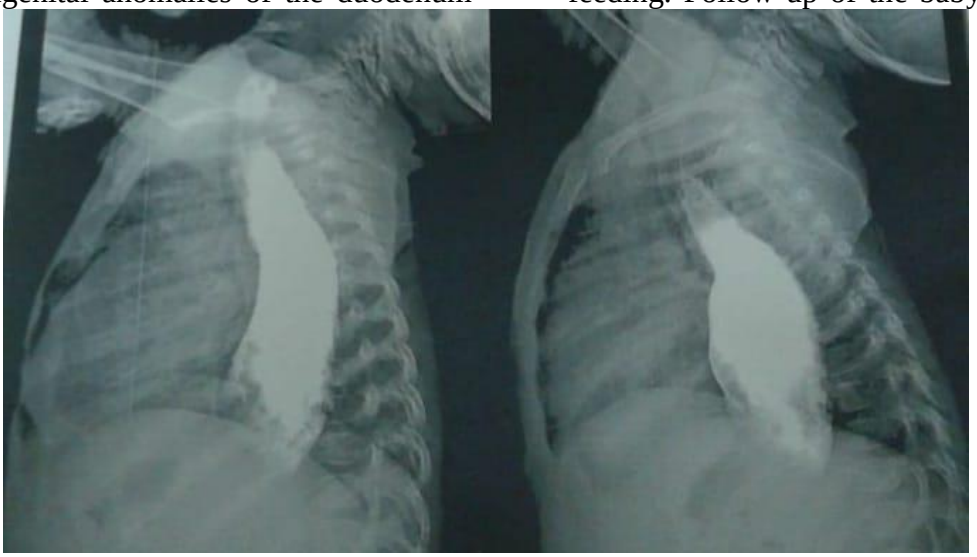

Figure 1. X-ray with barium meal contrast (upper GIT). It shows mega-esophagus with retained contrast and smooth taperin

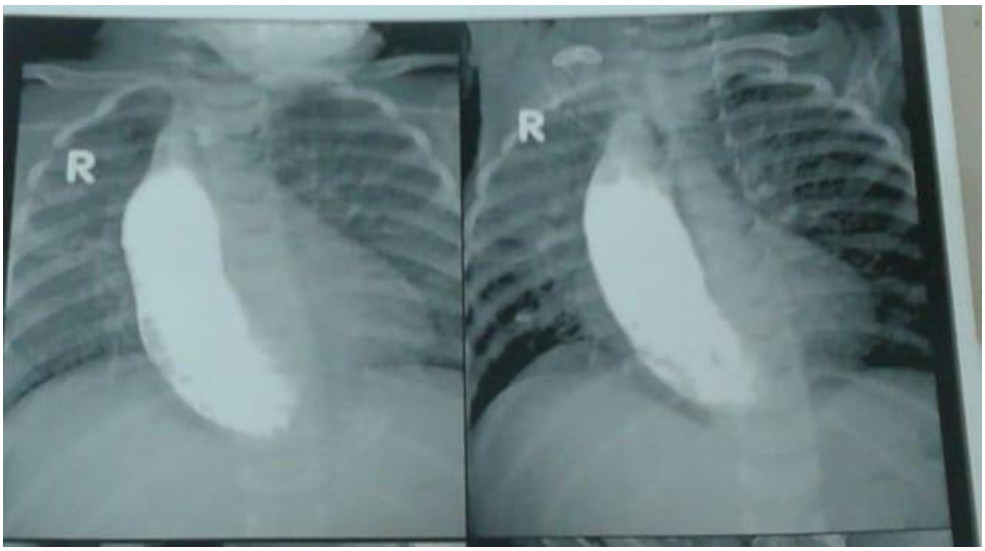

Figure 2. X-ray with contrast (lateral view) showing dilated esophagus.g at the distal end of esophagus (bird's beak appearance).

DISCUSSION

Cardiac achalasia is not common to occur in children, especially infants. Because of the rarity of the disease in infants, delayed diagnosis and treatment usually occurred [10]. King in 1953 reported the first case in infants. The infant was diagnosed at age of 6 months. Medical treatment failed and Heller's

esophagocardiomyotomy was performed after 3 months which lead to symptomatic improvement [2].

Because Cardiac achalasia is not common in infancy, we report this case and also highlight the difficulty of diagnosis. The main symptoms of cardiac achalasia are retrosternal pain and dysphagia which can't be informed by a non-verbal infant. Only adolescents and older children can describe these symptoms in detail [10]. Expulsion is the main symptom. Parents do not know the difference between vomiting and expulsion and insist that the act of expulsion was vomiting. Nevertheless, patients having achalasia may vomit, especially when complications as a huge mega-esophagus as, in such condition, occurred. Vomiting is initiated by the contraction of the lower end of the esophagus [12].

To detect the prevalence of cardiac achalasia in pediatrics, a global survey was done. Myers NA et al. Reported that the age of onset of symptoms had a wide distribution. 18% of patients begin symptoms in infancy and only 6% of patients could be diagnosed as cardiac achalasia. The disease is progressive so the symptoms may not appear during early childhood [13].

The gradual degeneration of the myenteric plexus of the esophagus gets worse as aging progresses, so the manifestation of the disease occurs during adolescence or adult life [6]. In contrast, regurgitation may occur in newborns due to cardiac achalasia but it is a very rare condition. This condition may be misdiagnosed and under-evaluated until the symptoms persist for long or become severe enough to affect infant's growth [14].

In our case, delayed diagnosis of cardiac achalasia was made at age of 11 months although vomiting is presented in the neonatal period. Much attention had been paid when vomiting persisted and led to recurrent RTI and failure to thrive. Also, Chatterjee et al [13], Choudhury et al [15], Shettihalli et al [16] and Iacob et al [17] reported delayed diagnosis of the cases because cardiac achalasia is a rare disease in infancy, and vomiting was misdiagnosed as gastroesophageal reflux.

Failure to thrive is a common associated disorder with cardiac achalasia. Although the age of our patient was 11 months, his weight was 6.5 kg (below the third centile) also Chatterjee et al [13] reported two cases of cardiac achalasia, one aged 9 months old, and his weight was 2.4 kg and the second was 11-months-old and his weight was 2 kg only. Also, Choudhury et al [15] reported an infant aged 7 months, and his weight was 2 kg. Failure to thrive occurred due to repeated vomiting of uncurdled milk and recurrent respiratory tract infection.

Keeping cardiac achalasia in mind in the diagnosis of the causes of intractable vomiting was suggested by Ashraf m who did a study at the pediatric department from Jan 2003 to Dec 2007 and reported ten children were diagnosed with cardiac achalasia, eight of them were infants [18].

This case was diagnosed by barium swallow with cineesophagogram showing dilatation of the esophagus, and smooth tapering of its lower end ("bird-beak" sign). The endoscopic assessment had been performed to detect the cause of obstruction and exclude other causes e.g., congenital membrane and acquired stricture. Endoscopy demonstrates retained food debris in the lower part of the esophagus. Although Ashraf et al [18] used esophageal manometry for diagnosis, we can't use it due to technical difficulties in children.

Medical treatment (isosorbide dinitrate or botulinum toxin injection) is used mainly in adults or older children. It provides a varying degree of success and short-term relief of symptoms. It could be given to patients having surgical risk factors. Dilatation of the esophagus using pneumatic dilators hasn't been used in infants to avoid many risks due to its disadvantages [19].

Esophagocardiomyotomy causes relief of dysphagia in 80% of cases for 5 years after operation and also decreases morbidity [19]. We referred the case to the surgical unit where a modified Heller's esophagomyotomy was performed.

The gold standard operation for relieving the non-organic obstruction of the lower end of the esophagus is Modified Heller's esophagocardiomyotomy with fundoplication either using open or laparoscopic approaches. This operation is safe and has good long-term results [21].

Chatterjee et al [13], Choudhury et al [15], Shettihalli et al [16] and Iacob et al [17] reported treatment of their cases by Heller's esophagocardiomyotomy operation with successful results.

After the operation, most children are relieved from manifestations and gained weight. But a few patients may still have mild dysphagia and may require drinking water during meals. About 5% to 15 % of cases may get complicated by squamous cell carcinoma mostly after 30 years so the following up is mandatory [22]. After the operation, our patient had a dramatic relief of symptoms, satisfactory weight gain, and the disappearance of pulmonary symptoms.

CONCLUSION

Cardiac achalasia is regarded as a rare disease in infants. Misdiagnosis as GERD is common. We should suspect this disease, especially if the infant has recurrent RTI, persistent vomiting and failure to thrive. An early diagnosis of these cases is needed for early and prompt surgical intervention for a successful outcome. It is better to do Heller's myotomy and do long-term follow-up due to the high risk of malignancy.

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