

## Early Childhood Achalasia Cardia: a local experience

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### ABSTRACT

**Introduction:** Achalasia cardia is rare in early childhood, presenting with difficulty in swallowing, regurgitation, choking, apnea, pneumonitis and impaired growth. Diagnosis depends on radiology and manometry. However; Clinical suspicion remains the key to diagnosis. Heller cardiomyotomy is safe and effective treatment.

**Methods:** Ten children with achalasia cardia, four of them infants, were seen over a 6-year period from Jan. 2002 to Dec. 2007. Male to female ratio was 3:2. Two of the affected children also had alacrima. Vomiting, cough, chest infection and failure to thrive were the presenting complaints and diagnosis was confirmed by barium study. All children underwent transabdominal modified heller esophagocardiomyotomy with anterior fundoplication.

**Results:** The most troubling symptom of vomiting settled after surgery and children took feed normally. There was no postoperative mortality and follow up showed excellent results.

**Conclusion:** Achalasia is rare in infancy. In our society, where consanguineous marriages are a tradition, physician should have high index of suspicion in children presenting with recurrent chest infections. The possible association of dry eyes with achalasia should also be considered. The best way to treat this malady is transabdominal modified heller's cardiomyotomy with anterior fundoplication.

**Keywords:** Achalasia cardia, alacrima, modified Heller cardiomyotomy

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### INTRODUCTION

Achalasia cardia is a neuromuscular disorder of unknown etiology, characterized by ineffective peristalsis in the distal esophagus and lack of receptive relaxation of lower esophageal sphincter (LES)<sup>1,2</sup>. Histopathologic findings include absence or deficiency of ganglion cells in the Auerbach plexus of the distal esophagus. Esophageal achalasia presents with difficulty in swallowing, regurgitation, choking, apnea, pneumonitis and impaired growth<sup>3,4</sup>. In infants, careful history will elicit that the vomitus characteristically contain uncurdled milk. Infants may predominantly have respiratory symptoms which may overshadow the vomiting. Diagnosis is mainly radiological with "Bird's beak" appearance and "megaesophagus" as typical findings. Because of its high sensitivity, manometry is the gold standard test for establishing the diagnosis. However, Clinical suspicion remains the key to diagnosis.

Achalasia is uncommon in childhood, and rare in infancy, reports about this disease are limited because of its extraordinary low incidence in

children<sup>5</sup>. The estimated incidence is 0.11 per 100 000 children<sup>6</sup> with less than 5% of patients under 4 years of age<sup>7,8</sup>. Disease involving many children in the same family may suggest familial or congenital etiology<sup>9,10</sup>. An association of adrenocortical insufficiency and alacrima(defective tear production) with achalasia has been termed Allgrove syndrome or "AAA" syndrome. This has been shown to affect siblings<sup>11</sup> and first cousins<sup>12</sup>. Allgrove syndrome is an autosomal recessive condition which was first described in 1978<sup>13</sup>. However, Haverkamp<sup>14</sup> et al. have reported combination of achalasia and alacrima in two brothers and a sister suggesting a separate entity of "AA" (double A) syndrome from well-described "AAA" syndrome.

Therapeutic options include medications, pneumatic dilatation and surgical myotomy. Drugs like nifedipine and isosorbidedinitrate reduce LES pressure. Botulinum toxin (Botox) may be injected into the lower esophageal sphincter to paralyze the muscles holding it shut. Balloon (pneumatic) dilatation stretches the muscle fibres of the lower esophageal sphincter forcefully. Heller's esophagocardiomyotomy invented in 1913<sup>15</sup> was popularized by Zaaier around 1923<sup>16</sup>. The treatment has a high degree of safety, effective results, and a long effective period<sup>17,18</sup>. It has been a standard surgical treatment for achalasia in North America and Europe in the recent years. The Heller myotomy is a lengthwise cut along the esophagus, starting above

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the LES and extending down onto the stomach. A partial fundoplication or "wrap" is generally added in order to prevent excessive reflux, which can cause serious damage to the esophagus over time.

**MATERIAL AND METHODS**

Ten children were diagnosed as achalasia cardia at Pediatric department of Fauji Foundation Hospital, Rawalpindi from Jan 2003 to Dec 2007. Male to female ratio was 3:2. Eight of them were infants and two were 4 years old. Two of the affected children also had alacrima. Children operated in other hospitals for the same condition and admitted only for nursing care were not included in study. Diagnosis of Achalasia was based on history, plain chest x-ray and barium study. The main symptoms were vomiting, cough, regurgitation and recurrent chest infection. Plain radiograph features included the absence of the fundic air bubble and presence of mediastinal air fluid level. The diagnosis was confirmed by a Barium cine-oesophagogram which showed typical esophageal dilation and a "bird beak" at the cardia. Manometry was not performed simply because of lack of this facility at the institution at the time of study. After diagnosis, children were shifted to surgical unit for further management. All patients underwent standard transabdominal esophagocardiomyotomy with anterior fundoplication.

**RESULTS**

Oral feeding was started after four hours in 8 infants and after 12 hours in older children. The most troubling symptom of vomiting settled after surgery and children took feed normally. Post operative gastro griffin swallow showed free flow of dye into the stomach, however, esophageal dilatation persisted. There was no post operative death. On three months follow up, children became asymptomatic, there was no complaint of reflux or regurgitation and all started gaining weight.

Table 1: Symptoms at presentation

Symptoms	n
Vomiting	10
Failure to thrive	10
Recurrent cough with fever	6
Recurrent respiratory distress	2
Alacrima	2

**DISCUSSION**

Esophageal achalasia was first described in 1674 by Thomas willis<sup>19</sup>. Shah has reported the incidence of 0.6 to 1 per 100,000 per year<sup>20</sup>. Afshan Fayyaz has reported incidence in children below 14 years of age as 1 case per 100,000 populations<sup>21</sup>.

Achalsiacardia is known to be a familial disease in children of consanguineous parents<sup>20</sup>. Afshan Fayyaz has also highlighted parental consanguinity.<sup>21</sup> In our country consanguineous marriages have solid tradition; therefore the recessive pattern of inheritance with greater chance of expression among their children, should be kept in mind.

In our study, two patients (one brother and his sister) had achalasia and alacrima. Therefore we had one family having children with "double A" syndrome. "Triple A" syndrome is very rare but "double A" seems fairly common. In our study male to female ratio is 3:2 with a predominantly infantile onset. Shah et al also reported that boys are affected more commonly than girls<sup>20</sup>. However, R. J. Thomas has given 3:4 male to female ratio<sup>22</sup>.

The gold standard for the diagnosis of achalasia is esophageal manometry. However, the paucity of availability of the facility and the cumbersome nature of the procedure, were the main reasons for us to rely on barium study as the tool of diagnosis. The same has been utilized by R.J. Thomas and et al<sup>22</sup>.

The objective of treatment of achalasia is to relieve functional obstruction in the distal oesophagus and esophagogastric junction. A modified Heller esophagocardiomyotomy is considered gold standard in the management of achalasia cardia<sup>16, 23</sup>.

A trans-abdominal oesophagocardiomyotomy with partial fundoplication was found to be the preferred mode of treatment with 98% resolution of symptoms<sup>22</sup>.

Perusal of literature revealed that clinical response to transthoracic esophagocardiomyotomy was fair, but good to excellent result were seen with transabdominal esophagocardiomyotomy<sup>24</sup>. This procedure when carried out laposcopically is safe and effective in children<sup>25</sup>. In our experience, abdominal esophagocardiomyotomy with anterior fundoplication gave excellent results.

In a worldwide survey of 175 paediatric cases of achalasia cardia, although 18% of patients had symptom onset during infancy, only 6% of the patients were identified as having achalasia during

infancy<sup>24</sup>. In view of the rarity of achalasia in infants, physicians are required to have high index of suspicion. Achalasia cardia should be kept in mind in the differential diagnosis of intractable regurgitation and dysphagia in infants. An early diagnosis would result in prompt surgical relief.

## CONCLUSION

Achalasia cardia is rare in children and infants, usually presenting with persistent vomiting, recurrent chest infections and failure to thrive. In our society, where consanguineous marriages are a tradition, physician should have high index of suspicion in children presenting with recurrent chest infections. The possible association of dry eyes with achalasia should also be considered. The best way to treat this malady is transabdominal modified heller'scardiomyotomy with anterior fundoplication.

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