

## Case Report

# Familial esophageal achalasia in mother and son

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### ABSTRACT

Familial esophageal achalasia a rare presentation, is most often described in monozygotic twins or siblings. Parent/child association is very rare and exhaustive review of the literature found about 5 times as many instances of affected siblings than instances of affected parent/child. Only seven instances of parent/child association have been reported till date. We report a 75 -year-old lady with achalasia cardia and her son who developed the same illness one year later. Mother was treated by Heller's surgery and son is presently on medical management.

**Keywords:** Achalasia, Esophagus, Familial

### INTRODUCTION

Familial esophageal achalasia a rare presentation, is most often described in monozygotic twins or siblings. Parent/child association is very rare and exhaustive review of the literature found about 5 times as many instances of affected siblings than instances of affected parent/child. Only seven instances of parent/child association have been reported till date.

We report a 75 -year-old lady with achalasia cardia and her son who developed the same illness one year later.

### CASE REPORT

A 75 year old lady without previous history of any systemic illness presented with dysphagia, retrosternal discomfort and vomiting of 10 days duration. She gave history mild dysphagia for the last 35 years which was non progressive and did not require any treatment. Now

for the last 10 days she had progressive dysphagia both for solids and liquids. This was associated with several episodes of vomiting. The vomitus contained foul smelling food particles. There was no hematemesis. She was on supportive treatment with intravenous fluids in a local hospital before she presented to us.

Her routine investigations, electrolytes, renal and liver function tests were within normal limits. Her barium swallow showed moderate dilatation with a typical parrot's beak appearance of the lower end of the esophagus suggestive of achalasia cardia.

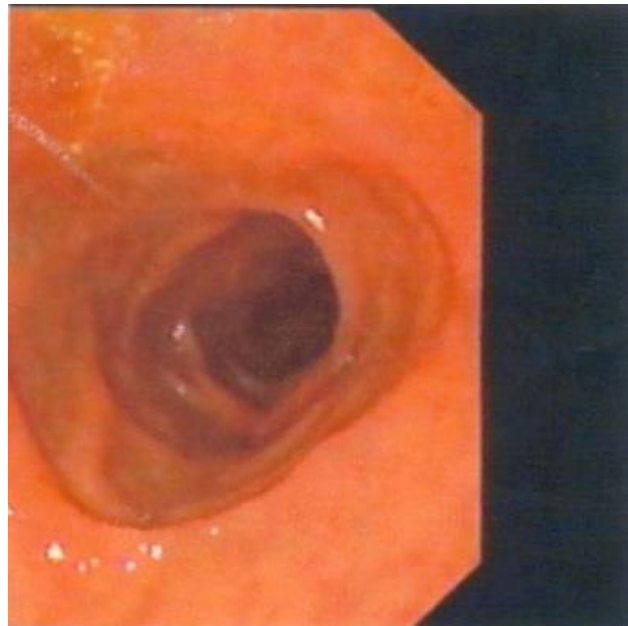
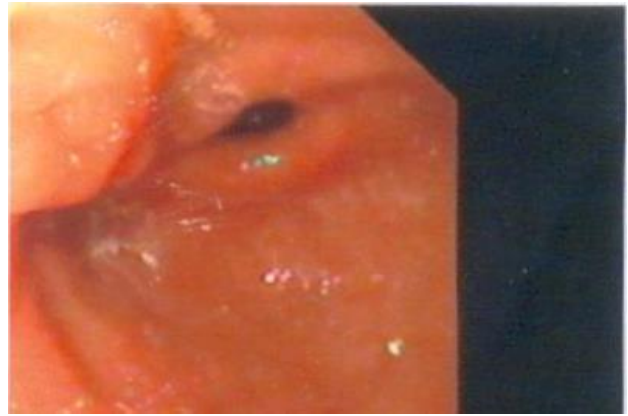
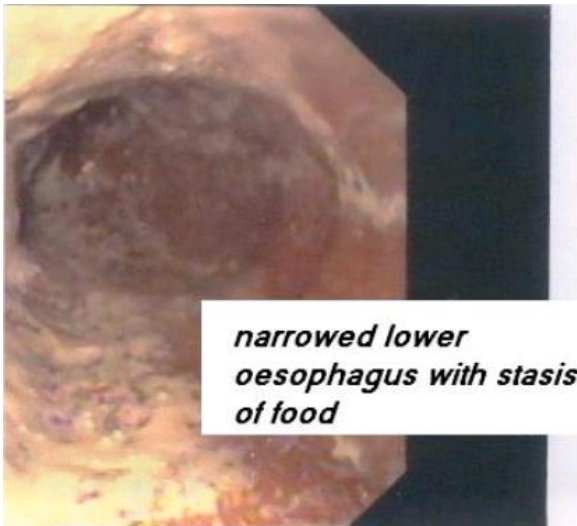
Her upper gastrointestinal endoscopy revealed dilated esophagus in the middle and lower third, with a narrowed lower end, with stasis and no detectable growth.

She was treated with nitrates and soft foods initially but medical treatment proving ineffective was subjected to Heller's extra mucosal myotomy of the Lower

oesophageal sphincter. Her symptoms improved and she has been doing well till date.



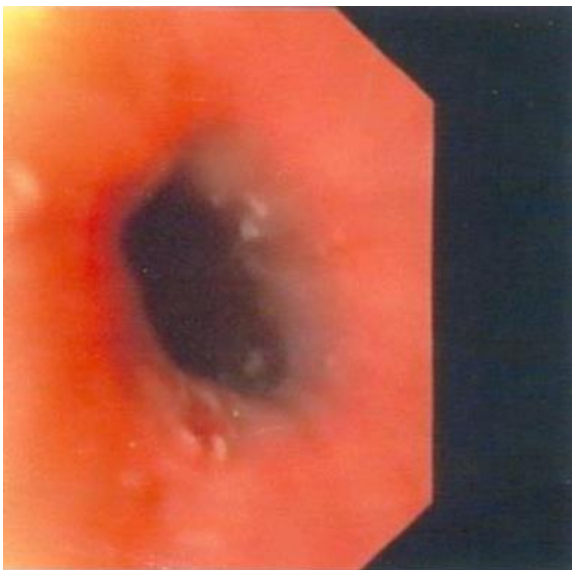
**Figure 1: Parrot's beaking at the lower end of oesophagus.**



**Figure 2: Narrowed lower end of oesophagus with stasis of food.**

One and a half years later, her son a 48 year old man born of non consanguineous marriage, presented with complaints of dysphagia to solids for 6 months and regurgitation of food. His barium swallow revealed a dilated esophagus with narrowing at the lower end. Upper gastrointestinal endoscopy revealed a dilated esophagus with a slightly narrowed lower part and there was no growth seen at the cardia.

He is being treated with Isosorbide dinitrate 10mg twice daily orally and currently responding well on medical management. He is currently on follow up for the last six months and is placed on continued follow up. Oesophageal manometry was done on follow up which showed normal lower oesophageal sphincter pressure. There was reduction in the duration of swallow induced relaxation. No other family member had similar symptoms.



## DISCUSSION

Achalasia is a motor disorder of the esophageal smooth muscle in which the lower esophageal sphincter does not relax normally with swallowing. Primary idiopathic achalasia accounts for most of the patients. Secondary achalasia may be caused by gastric carcinoma that infiltrates the esophagus, lymphoma, Chagas' disease, certain viral infections, eosinophilic gastroenteritis, and neurodegenerative disorders.

The aetiopathogenesis of primary achalasia is largely unknown, although an immunogenetic predisposition is suspected. Familial occurrence of achalasia is rare.<sup>1</sup> Thibert et al (1965) described 2 families, each with 2 affected siblings.<sup>4</sup> Frieling et al (1988) described 4 families with multiple cases.<sup>2,3</sup> Family occurrence of esophageal motor disorders supports the hypothesis that a genetic trait may play a role in the pathogenesis.

Familial achalasia is extraordinarily rare, for an exhaustive search of the world literature yielded only about two dozen cases. Most of these cases occur among siblings and familial achalasia is likely transmitted as an autosomal recessive trait.<sup>7</sup> A case of familial achalasia was described in an 8-month old Sudanese male child in 2004.<sup>10</sup>

Vertical transmission, that is disease manifestation in parent and child is very rare suggesting a common environmental factor or an autoimmune etiology. Further studies need to be done to prove this etiology.<sup>5,8</sup> The triple A syndrome, also known as Allgrove syndrome<sup>6,9</sup>, recessively inherited due to a malfunction in chromosome 12q 13, is characterized by the clinical triad of adrenocorticotrophic hormone (ACTH)-resistant adrenal failure, achalasia cardia and alacrima. The syndrome is

associated with variable and progressive neurological impairment, dermatological features such as palmoplantar hyperkeratosis, other signs including short stature, osteoporosis and microcephaly.

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