Familial Achalasia Cardia in Childhood: A Case Report of Three Siblings

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ABSTRACT

Achalasia cardia is a rare condition in the paediatric population, and familial occurrences are even rarer. The authors herein report a case of three siblings diagnosed with familial achalasia cardia. The first case was a 12-year male who presented with progressive dysphagia to solids and later to liquids for one year. A diagnosis of achalasia cardia was made based on a barium swallow and meal, and he underwent Heller's myotomy with an uneventful recovery. His eight-year sister also presented with similar symptoms, including progressive dysphagia and intermittent non-bilious vomiting for one year. She was diagnosed with achalasia cardia, underwent Heller's myotomy, and had a smooth postoperative course. At two years of follow-up, both siblings were symptom-free and thriving. The third sibling, a four-year, developed similar symptoms and was diagnosed with achalasia cardia six months ago. He also underwent Heller's myotomy. This report highlights the rare occurrence of familial achalasia cardia in three siblings and underscores the potential genetic predisposition in the pathogenesis of this condition.

Key Words: *Achalasia cardia, Familial, Paediatrics*.

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INTRODUCTION

Achalasia is characterised by functional obstruction of the distal oesophagus due to failure of relaxation of the lower oesophageal sphincter and loss of oesophageal peristalsis.[1]

Achalasia is a rare diagnosis in children. Its estimated incidence is approximately 0.11 to 0.18/100,000 children per year. It can present as early as the 7th week of age, but its incidence under the age of 15 years is 5%.[1,2] Its familial presentation is extremely rare; it mostly occurs among siblings or in monozygotic twins.[2] Familial achalasia occurring in mother and daughter in adult life is also reported in the literature.[1] The authors report these cases of familial achalasia cardia that is extremely rare and can be successfully treated with surgical management.

CASE REPORT

A 12-year male and his eight-year sister presented to the outpatient department with complaints of progressive dysphagia, initially to solids and then to liquids for one year. They were investigated, and on barium swallow, both cases were suggestive of achalasia cardia, showing mildly dilated oesophagus with significant hold-up of contrast at its distal end and smooth narrowing, but the contrast passed into the stomach.

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Heller's myotomy was performed in both cases, and the postoperative period was uneventful. Both patients were discharged on the 4th postoperative day. The young girl also had a hearing impairment, for which an ENT consultation was sought. After two years of follow-up, both siblings were symptom-free and thriving well. However, their 3rd sibling, aged four years, developed similar symptoms and was diagnosed with achalasia cardia six months ago. He underwent Heller's myotomy at another hospital.

DISCUSSION

Achalasia cardia was first described by Thomas Williams in 1674. The underlying aetiology of achalasia cardia remains unknown. Genetic predisposition is considered one of the possible contributing factors to the pathogenesis of achalasia cardia.[1] Less than 1% of achalasia cardia cases are seen as isolated familial patterns. Familial cases mostly show horizontal transmission. The mean age at diagnosis is usually 8.8 years.[2] However, Bosher and Shaw reported two siblings with achalasia cardia in early infancy.[3] It usually presents with feeding problems, failure to thrive, and repeated respiratory tract infections. In the present patients, it presented with progressive dysphagia and non-bilious vomiting.

Achalasia cardia mostly presents alone; however, various syndromes are also associated with achalasia, such as Down's syndrome, Allgrove's syndrome, and Sjogren's syndrome.[4] Tebaibia *et al*. reported 18 familial cases of achalasia cardia. In their study, 18 families with familial achalasia were identified (n = 41 patients). Two members were affected in each of 14 families, three members in each of three families, and four affected members in one family. Consanguinity was found in 89% of

patients, and parent-to-child vertical transmission was found in all cases. Achalasia was associated with Allgrove syndrome in 15 families and was isolated in three families. There was no difference between the two groups for age, age at onset, gender, and the presence of the cardinal signs of achalasia.[5]

Barium swallow and meal, esophagoscopy, and manometry are needed to diagnose achalasia.[6] Detailed history and physical examination are essential for accurate diagnosis of achalasia cardia in children.[7] Barium swallow and meal show dilatation of the proximal oesophagus with a "bird's beak" or "rat tail" sign, indicating narrowing of the distal end due to the contracted lower oesophageal sphincter.[6] Similar findings were present in the present case.

The differential diagnosis of achalasia cardia in children includes other potential causes of dysphagia.[6] Upper gastrointestinal endoscopy can be used to rule out other diagnoses. Oesophageal manometry remains the most reliable technique for diagnosing achalasia. It may demonstrate variable lower oesophageal sphincter resting pressures.[6-8] However, manometry is not routinely used in the paediatric population, as it requires a large catheter probe and may have poor tolerance. The use of sedatives during the procedure can also interfere with findings.[9]

There are both medical and surgical options for treating achalasia. While studies on the effectiveness of calcium channel blockers in adult patients exist, research on their impact in paediatric cases is limited. Botulinum toxin injection is effective in both paediatric and adult populations. However, balloon dilation (BD) or Heller's myotomy is considered the definitive treatment.[10] BD is effective in adults, but studies have shown that Heller's myotomy (open/laparoscopic) is the most definitive treatment option for paediatric achalasia.[10] Recent advances in the treatment of achalasia include per-oral endoscopic myotomy (POEM). While POEM is a promising therapeutic approach, its use is restricted by the lack of an antireflux procedure, which may increase the chances of gastroesophageal reflux. Early outcomes from POEM are encouraging, but larger sample sizes and long-term outcomes are needed to validate its use as an alternative treatment option.[11] In conclusion, this report highlights the rare occurrence of familial achalasia cardia in three siblings and underscores the potential genetic contribution to the pathogenesis of this condition in familial cases.

PATIENTS' CONSENT:
Informed consent was obtained from the parents of the patients for publication of this case report.

COMPETING INTEREST:
The authors declared no conflict of interest.

AUTHORS' CONTRIBUTION:
HM: Manuscript drafting and literature search.
SIN: Case selection and manuscript editing.
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NT: Manuscript editing and final approval.
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