

Achalasia cardia in children: A report of two cases

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Oesophageal achalasia is a neuromuscular disorder of unknown aetiology, characterised by abnormal motility of the oesophagus and failure of the lower oesophageal sphincter to relax. This causes an abnormal dilatation of the oesophagus and resultant symptoms of vomiting/regurgitation, dysphagia, chest pain and at times, signs of lung aspiration and infection. In children, it may present as a chronic cough. The condition usually presents in the 4th and 5th decades and has very rarely been described in children. We describe two cases of achalasia and their imaging findings in adolescents.

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Case report

Case 1

A 13-year-old boy presented to the surgical outpatient department with a history of intermittent vomiting and dysphagia since the age of 3 months. Clinically, he was cachectic with no syndromic features. On oesophagogastrosocopy, the oesophagus was markedly dilated, with food residue and a spastic and stiff lower oesophageal sphincter (LES). No fibrotic rings were identified and there were no signs of oesophagitis. The opening was less than 3 mm and it was not possible to pass the scope beyond the stiff LES; hence, manometric studies were not done.

A control chest radiograph (Fig. 1A) showed a massively dilated oesophagus with an air-fluid level. A subsequent barium swallow study (Fig. 1B) demonstrated a mega-oesophagus, with irregular and abnormal peristalsis in the proximal oesophagus. The oesophagus tapered and narrowed at the gastro-oesophageal junction, with failure of the LES to relax. No aspiration changes were noted in the lung fields. A diagnosis of achalasia was made and the patient was booked for gastroscopy and pneumatic dilatation. The LES was dilated up to 14 mm without complications. The patient was symptom free and was discharged, and requested to come back for a review examination after 6 months. The patient presented again after 3 months with symptoms similar to initial presentation, and a repeat barium swallow again showed a tight stricture at the distal oesophagus, suggesting recurrence of achalasia. As the symptoms recurred within 6 months of dilatation, and as the patient was otherwise young and healthy, surgery was chosen as a treatment of choice as it provides lasting benefits for children with achalasia. Medical treatment with Botox (botulinium) injections was not

selected because it needs to be repeated multiple times a year with a mean duration of symptom relief of about 4 months. A Heller's myotomy and fundoplication were done without any complications. A follow-up examination after 2 months and then 1 year showed complete resolution in clinical symptoms with no signs of recurrence to date.

Case 2

A female patient with juvenile diabetes mellitus and hypogonadism presented with dysphagia and vomiting at the age of 16 years. A barium swallow study confirmed the findings of achalasia (Figs 2A and B). There were no signs of hyperpigmentation or alacrimia to suggest triple AAA syndrome (achalasia, alacrimia, adrenocorticotrophic hormone (ACTH) deficiency). Manometric studies confirmed high LES pressure (47.1 mmHg) and a poor relaxation of 30.4%. Medical treatment was not considered as the results have either a short-term success or are not well studied in children. She was treated with Heller's myotomy, which was complicated by perforation and later severe peritoneal sepsis. The patient subsequently had a long admission to the intensive care unit followed by a high care unit, but despite aggressive therapy for the sepsis in the context of her diabetes mellitus, she died 9 months after the procedure.

Discussion

Achalasia cardia is a neuromuscular disorder of unknown aetiology, rarely described in children and adolescents. The symptoms include dysphagia, vomiting/regurgitation of food, retrosternal pain, poor growth and respiratory symptoms due to chronic aspiration.^[1-3] Few cases have been reported in infants, and some familial forms have been described but are even more rare.^[4,5] An unusual case of achalasia in

a child presenting with chronic cough due to compression and narrowing of the trachea has been described.^[6]

A postulated cause of achalasia is the degeneration of the ganglion cells in Auerbach's plexus of the oesophagus. The

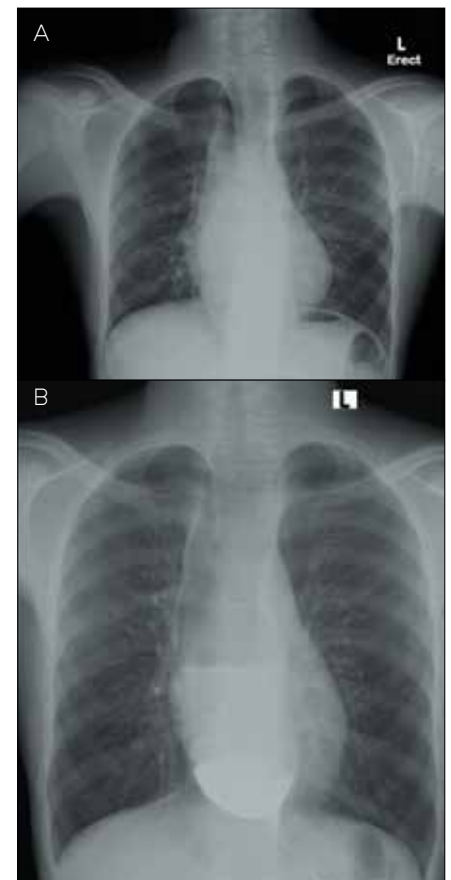


Fig. 1. A. Control anteroposterior demonstrating a massively dilated oesophagus with an air-fluid level; B. Lateral barium swallow radiograph confirming a mega-oesophagus with air contrast level and a classic 'bird beak' appearance of the distal oesophagus of the patient described in Case 1.

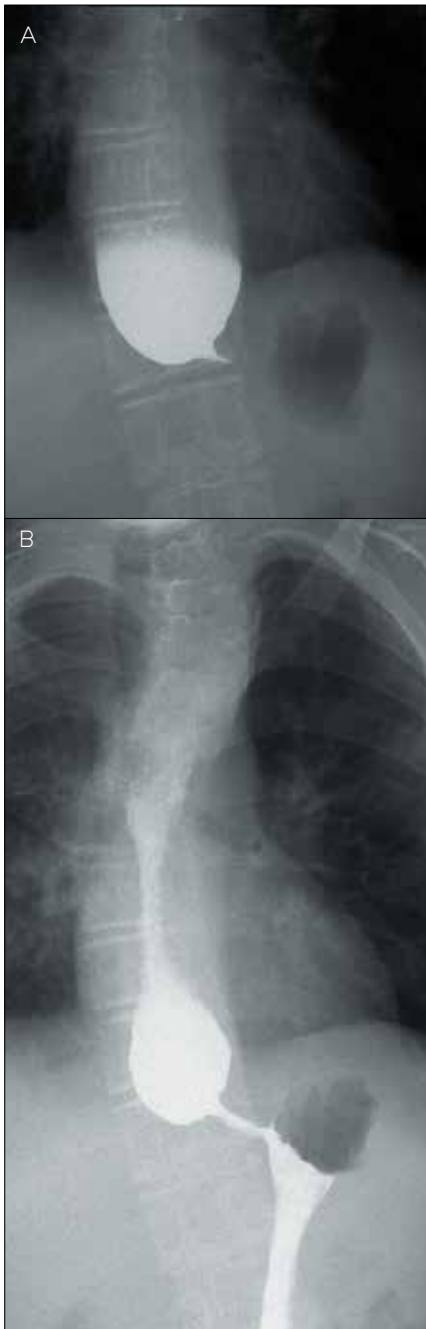


Fig. 2. Frontal barium swallow images demonstrating: A. a dilated oesophagus with a contrast fluid level in the superior mediastinum; B. a classic 'bird beak' appearance of the distal oesophagus in the patient described in Case 2.

result is a lack of relaxation of the LES, with dilatation and hypertrophy of the proximal portion of the oesophagus and disordered peristalsis in the rest of the oesophagus.^[1,3,7] Numerous syndromes have been associated

with achalasia, including AAA. Other associations include progressive cerebellar ataxia, parkinsonism, familial glucocorticoid deficiency, mental retardation and Down syndrome. The steroid abnormality of AAA is often not appreciated and may appear as hyperpigmentation or hypoglycaemia-induced symptoms of dizziness or confusion.^[2-5,8]

The gold standard for diagnosis of achalasia is a barium swallow and manometric studies. A barium swallow demonstrates the presence of mega-oesophagus and the so-called 'bird beak' tapering of the lower oesophagus, causing functional obstruction and significant delay of passage of contrast into the stomach. Non-peristaltic tertiary contractions may also be seen.^[6,8,9]

At endoscopy, a dilated oesophagus with a tight LES that 'pops' open with gentle pressure is often observed, as well as retained food and saliva. Signs of oesophagitis may also be seen. A normal oesophagogastroscope does not exclude the diagnosis, as 40% of endoscopies may be normal.^[3,8,9] Oesophageal manometry defines achalasia as the absence of peristalsis or the presence of abnormal peristalsis in the oesophageal body, a stiff LES with a resting pressure of >45 mmHg with poor or incomplete relaxation, and a residual pressure after emptying of >8 mmHg.^[3,7,9] Biopsy, if performed, may show lack of myenteric plexus innervation.

Treatment options depend on a patient's willingness to undergo an invasive procedure and on their physical ability to endure it. The medical treatment includes calcium channel blockers such as nifedipine, which inhibits the transmembrane influx of calcium in cardiac and smooth muscle; this has been used primarily in adults to treat achalasia. The use of calcium channel blockers in children, however, is not well studied. Other medical treatment options include endoscopic injection of Botox into the LES. Botox acts on the terminal nerve endings, preventing the release of acetylcholine. This can be both diagnostic and therapeutic; however, in children the optimal dose and injection frequency to relieve achalasia is not determined and the duration of symptom relief is only 4 months, thus often requiring multiple treatments. Pneumatic dilatations have been used in children with a functionally obstructed oesophagus. Multiple dilatations are often required to achieve complete relief of symptoms and can be complicated by

substernal pain, perforation and gastro-oesophageal reflux disease. Surgery is the most definite treatment of choice, and includes Heller's myotomy with or without fundoplication. Two of the most important complications of surgery include perforation (as in Case 2) and recurrent dysphagia.^[1,3,8,9]

A newer, relatively less invasive technique called POEM (per oral endoscopic myotomy) has been used for the treatment of achalasia in adults, and directly treats the diseased tissue using an oral approach. POEM is safe and equally effective in children; however, it is not yet widely employed in the management of paediatric achalasia.^[1]

Conclusion

Achalasia cardia is a rare motor disorder in children, but should be kept in the differential diagnosis of patients with a history of intermittent chronic regurgitation of food and dysphagia. Alertness on the part of the paediatrician and radiologist with respect to the disease should lead to an early diagnosis and prompt, definite treatment. There are numerous treatment options available, and the choice depends mostly on willingness and compliance of the patient.

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