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Case 1: An adolescent with chronic cough and wheezing

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A 16-year-old-boy was evaluated for a two-year history of nocturnal cough and wheezing. He had not responsed to treatment with inhaled corticosteroids and long-acting $beta_2$-agonists, a proton pump inhibitor and several courses of azithromycin. His medical history revealed no seasonal or food allergies, recurrent infections, sinusitis or diarrhea. Allergy skin testing was negative. Additional history received with the referral noted the boy experienced difficulty swallowing saliva and weight loss of several kilograms during the year before the underlying diagnosis was eventually established. On physical examination, his heart rate, respiratory rate, oxygen saturation and chest auscultation findings were normal. Lung spirometry showed a mild obstructive pattern with flattening of the expiratory flow-volume loops (Figure 1), which remained unchanged after administering salbutamol. Because of his persistent symptoms, flexible bronchoscopy was performed, which identified a marked flattening of the lumen along the full length of the trachea. The cell count in the bronchoalveolar lavage fluid contained 20% neutrophils and 4% eosinophils, and staining with Oil red O was positive, indicating the presence of fat droplets. Subsequent bronchoalveolar lavage culture was negative. Further testing confirmed the diagnosis.

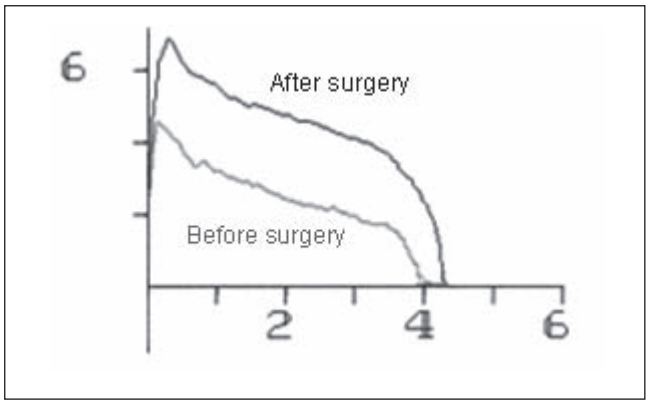

Figure 1) *Spirometry before and after surgery*

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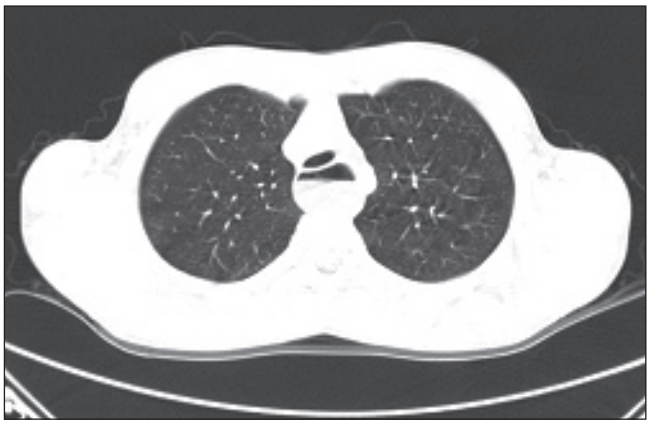

Figure 2) *Chest computed tomography scan showing dilation of the esophagus causing compression of the trachea*

CASE 1 DIAGNOSIS: ACHALASIA

Chest computed tomography (CT) scans revealed a megaesophagus (maximum axial diameters 50 mm × 35 mm), causing compression of the trachea, with a fluid level containing dense food residue, accumulating in the dilated distal esophagus (Figure 2). A barium swallow examination showed 'bird's beak' features (Figure 3) characteristic of achalasia. Achalasia is a primary esophageal motility disorder of uncertain etiology, which is rare in the paediatric age group, with an estimated overall annual incidence of 0.5 to one cases per 100,000 live births (1,2). Only 2% to 3% of all cases occur in childhood. It is characterized by the absence of peristaltic waves in the body of the esophagus and failure of the lower sphincter to relax in response to swallowing, resulting in an abnormal dilation of the esophagus that may also interfere with airway patency. Degeneration of the myenteric plexus and a consequent lack of the inhibitory neurons required for lower sphincter relaxation and peristaltic contraction are believed to be involved in the pathogenic mechanism (3). Achalasia usually presents with gastrointestinal symptoms including dysphagia, chest pain, vomiting and regurgitation of undigested food, often with consequent weight loss. Respiratory manifestations (chronic cough, especially at night or with recumbency, choking, recurrent pulmonary infections, pneumonia, wheezing, atelectasis and pulmonary empyema [4]) are less common but may be the presenting signs of this condition.

The diagnosis of achalasia can present a challenge when respiratory symptoms are the main presenting complaint. To avoid misdiagnosis and prevent the onset of complications, achalasia should be considered in the differential diagnosis of chronic cough, choking, recurrent pulmonary infections, pneumonia and wheezing. In such situations, the clinician should inquire about the presence of dysphagia, chest pain, regurgitation of food that is nonacidic and coughing or choking that worsens with recumbency. Achalasia-related lung disorders are mainly caused by fluids and food particles overflowing from the dilated esophagus into the trachea, causing aspiration and persistent upper airway inflammation. Chronic cough, dyspnea and stridor can also be due to compression of the trachea by the megaesophagus. In the present case, the morphology of the flow-volume curve suggested tracheal compression. He was investigated by flexible bronchoscopy, which confirmed a marked tracheal compression. The definitive diagnosis of achalasia was suggested by the CT scan and barium swallow examination, which demonstrated a megaesophagus. Manometry has been considered the gold standard for diagnosing achalasia but

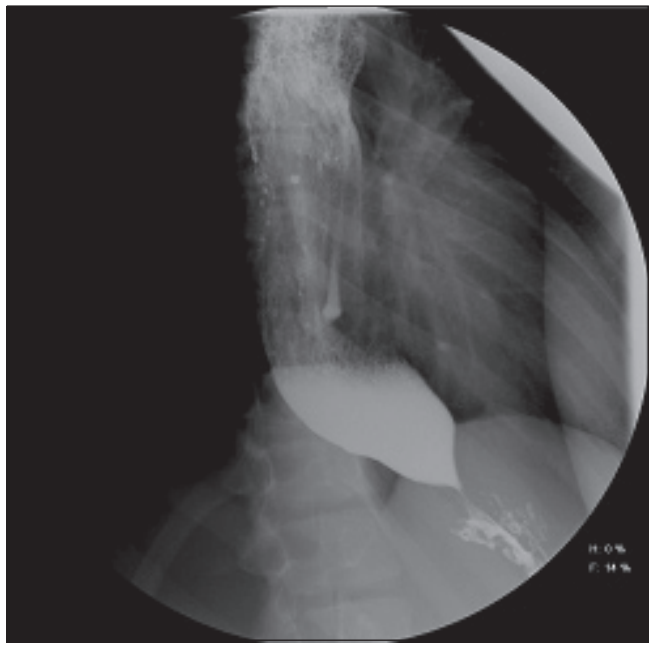

Figure 3) *Barium swallow showing typical 'bird's beak' features*

was not performed in the present case because the diagnosis was established on the basis of the CT scan and barium swallow examination. The boy underwent surgical correction with laparoscopic esophagomyotomy and partial anterior fundoplication (Heller-Dor procedure). During follow-up at four months after surgery, he reported no cough or any other symptoms and had gained 5 kg in weight. Spirometry showed improvements in all parameters (forced expiratory volume in 1 s was 113% of predicted, forced expiratory volume in 1 s/forced vital capacity was 95% of predicted, forced vital capacity was 107% of predicted, and forced expiratory flow between 25% and 75% of vital capacity was 119% of predicted) and the flow-volume curve had a normal morphology (Figure 1).

CLINICAL PEARLS

- While uncommon, a greater awareness of achalasia in childhood could facilitate earlier diagnosis, which may avoid unnecessary investigation and treatment and prevent the onset of complications.
- Achalasia should be considered in the differential diagnosis of chronic cough, choking, recurrent pulmonary infections, pneumonia and wheezing.
- CT scans and spirometry can act as complementary diagnostic tools in achalasia.

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