CASE REPORT

Oesophageal achalasia in an 11-year-old patient with persistent respiratory symptoms and suspected bronchial asthma

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ABSTRACT

Oesophageal achalasia is a disease characterized by the inability of the lower oesophageal sphincter (LOS) to relax, which results in persistent contraction and the lack of peristaltic movement in the oesophageal body. It is a rare condition in children; therefore, its identification may be challenging. In this paper, we present a case of an 11-year-old patient with persistent cough and post-exercise tachypnoea, consulted due to uncontrolled asthma. After thorough diagnostic work-up, the patient was given the final diagnosis of oesophageal achalasia and was referred for surgical treatment. Uncontrolled asthma always requires the supervision of inhalation technique, consideration of treatment modification, and confirmation of the diagnosis. In this case, in-depth anamnesis and adequate interpretation of lung function tests allowed a correct diagnosis.

KEY WORDS:
bronchial asthma, oesophageal achalasia, spirometry.

INTRODUCTION

Oesophageal achalasia is a rare oesophageal motility disorder characterized by the inability of the lower oesophageal sphincter (LOS) to relax, resulting in its persistent contraction, and the lack of peristaltic movement in the oesophageal body [1]. These lead to the slower passage of swallowed food and liquids, and oesophageal retention and inflammation [2]. The most common complaints include dysphagia, regurgitation, aspiration, chest pain, persistent cough, and weight loss [1]. Respiratory symptoms such as pneumonia, bronchiectasis, stridor, and acute respiratory insufficiency from tracheal compression by a severely dilated oesophagus may also occur [3]. Due to non-specific symptoms and very low incidence (approx. 1/100,000) in the paediatric population, the diagnosis is rarely considered among children [2]. Of note, only 5% of the cases occur under the age of 15 years [2].

Here, we present a case of an 11-year-old patient consulted for poorly controlled asthma, who was finally diagnosed with oesophageal achalasia.

CASE REPORT

An 11-year-old boy with chronic cough, no history of gastrointestinal symptoms, morning sputum expectoration and post-exercise tachypnoea for the last 2 years, a family history of bronchial asthma, and tobacco smoke exposure at home, was admitted to our department due to uncontrolled bronchial asthma. According to the boy’s mother, asthma was diagnosed 2 months earlier based on his history and wheezing on auscultation (no medical record available). Asthma treatment with twice-daily fluticasone 125 μg + salmeterol 25 μg was introduced with partial reduction of symptoms. Additionally, in the last 18 months he experienced 2 aspiration episodes – one

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during the induction of anaesthesia before a planned adenoidectomy, and one at home resulting in loss of consciousness and subsequent hospitalization.

On admission, he was in a good general condition. Physical examination revealed obesity (BMI 27.2 kg/m²; 98th percentile), noisy breathing, and discrete inspiratory wheezing over both lungs. Results of a basic metabolic panel, WBC with peripheral blood smear, and immunoglobulin levels (IgA, IgG, IgM, IgE) were within normal values. No atopy was detected to common inhalant allergens by allergen-specific IgE and skin prick tests. Spirometry revealed a lowered FEV1/FVC ratio, indicating an obstructive defect, and an increased FEV1/PEF ratio suggestive of upper/central airway obstruction. Bronchodilator reversibility test was negative (Figure 1). Electrocardiography and echocardiography were normal, and polysomnography showed a moderate obstructive sleep apnoea. During hospitalization, the boy expectorated significant amounts of secretion containing food particles, after which he revealed dysphagia and 4 kg weight loss over the past 2 months.

During induction to anaesthesia before bronchoscopy, another mild episode of aspiration took place. Flexible bronchoscopy revealed a ball-like protrusion in the posterior wall of the upper third of the trachea, with severe narrowing to periodic luminal collapse in the middle part of the trachea, indicating tracheal compression. Subsequently, an urgent gastroscopy was performed during the same anaesthesia showing a massively dilated lower oesophagus between distal 30 cm and 20 cm, suggestive of oesophageal achalasia. Upper gastrointestinal tract X-ray (Figure 2) confirmed the diagnosis, revealing dilation of the oesophagus along its whole length and delayed passage of contrast medium – after 20 minutes in an upright position it was still present in the lower part of the oesophagus.

Contrast-enhanced computed tomography of the chest showed dilated oesophagus filled with fluid on its entire length; the lumen diameter was 6 cm and wall thickness 3.5 mm. No pathological structures modelling the oesophagus were found (Figure 3). High-resolution manometry further confirmed the diagnosis – LOS resting pressure was 29 mm Hg (normal value 10–35 mm Hg) and integrated relaxation pressure (IRP) was 16.4 mm Hg (normal value < 13 mm Hg) [4]. Based on the lack of peristalsis, oesophageal achalasia subtype I was identified, and the boy was referred for surgical treatment. Following a laparoscopic Heller myotomy with Toupet fundoplication, the patient’s condition improved, and he was discharged home in good condition.

DISCUSSION

Chronic cough is one of the most common symptoms in children. The most prevalent causes include bronchial asthma, post-infectious cough, protracted bacterial bronchitis, airway or cardiological malformations, bronchiectasis, and postnasal drip [5, 6]. In school-age children, the most common reasons for chronic cough are asthma and asthma-like symptoms [7]. The diagnosis of asthma is based on clinical symptoms, lung function tests results, and response to treatment.

Our patient was initially diagnosed with bronchial asthma due to his history of persistent cough, post-exercise tachypnoea, wheezing on auscultation, and a positive family history of asthma. However, the response to treatment was only partial, and he did not present episodes
FIGURE 2. Upper gastrointestinal tract X-ray after the oral administration of an iodinated contrast medium (Visipaque). All photos show dilation of the oesophagus and deposition of contrast in the lower part of the oesophagus.

FIGURE 3. Computed tomography of the chest showing tracheal compression and dilation of the oesophagus, non-contrast projections (A, B), and contrast-enhanced projections (C, D).

* Oesophagus.
of wheezing or dyspnoea requiring bronchodilators, night symptoms, or atopy. Of note, during hospitalization he presented inspiratory wheezing typical of upper airway obstruction, while in asthma expiratory wheezing is the most frequent abnormality.

In cases of poor asthma control, the supervision of inhalation technique, consideration of treatment modification, and verification of the diagnosis must be performed. Our patient’s inhalation technique was assessed as correct. Before treatment step-up, the diagnosis of asthma was verified. While in spirometry our patient showed a lowered FEV1/FVC ratio with lowered FEV1, allowing the diagnosis of severe airway obstruction; it was not reversible after the administration of salbutamol. Of note, an increased FEV1/PEF ratio (0.62) was suggestive of upper/central airway obstruction [8, 9]. Assuming upper airway obstruction as a possible cause of the presented symptoms, videobronchoscopy was performed to exclude potential airway malformations, dysfunctional breathing, or inhaled foreign body. The study revealed tracheal stenosis, which was caused by oesophageal achalasia.

Tracheal stenosis in children is rare, in most cases primary, and generally congenital [10]. In primary tracheal stenosis the narrowing of the tracheal lumen results from the disease concerning the trachea, and in most paediatric cases it manifests as complete tracheal rings. Secondary stenosis occurs when other structures compress the trachea from the outside. These may include diseases affecting the thyroid, thoracic blood vessels, thymus, mediastinal lymph nodes or tumours, cysts and abscesses localized in the mediastinum or neck, and oesophagus [11]. In the presented case, these were rejected based on the results of echocardiography, electrocardiography, and contrast-enhanced chest CT scan.

The diagnosis of oesophageal achalasia was in our case hampered by a family history of asthma, partial, yet present improvement of symptoms after initiating asthma treatment, and obesity. Among patients with oesophageal achalasia 35% to 91% report weight loss during initial presentation [12–14], as did our patient. Interestingly, obese patients may have more frequent choking or vomiting symptoms, which was the case in our patient [14]. Although our patient lost weight (4 kg in 2 months), it was not reported on admission because his mother attributed it to the introduction of a new diet.

The diagnosis of oesophageal achalasia was based on gastroscopy, X-ray with iodinated contrast medium, and, preferably, high-resolution manometry (HRM) [1]. Classic endoscopic examination shows a dilated oesophageal body with a puckered LOS and proximally retained food and saliva. Typical findings in X-ray include the visualization of dilated oesophagus over time (bird beak sign) with corresponding delayed passage from the oesophagus to the stomach [15]. The gold standard diagnostic test for achalasia is HRM, which usually reveals a lack of peristalsis and incomplete LES relaxation with increased integrated relaxation pressure (IRP). An IRP value greater than 15 mmHg is strongly suggestive of achalasia [16]. HRM result allows the classification of achalasia into 3 distinct subtypes, with subtype II achalasia showing the best response to treatment and subtype III achalasia being the most difficult to treat [17, 18]. Our patient presented clinical features of achalasia in all the conducted above-mentioned examinations. He met the criteria for subtype I achalasia in HRM [14].

Available treatment options for achalasia include pharmacological, endoscopic (botulin toxin injection or oesophageal balloon dilation), and surgical treatment [14, 16]. Laparoscopic Heller myotomy with fundoplication is considered the most effective and was performed in our patient [2]. A more recent treatment method is peroral endoscopic myotomy (POEM), which gives similarly promising results but is a less invasive procedure [19].

CONCLUSIONS

Patients with asthma require regular assessment of inhaler technique and adherence. In case of unresponsive asthma, modification of treatment and verification of the diagnosis should be considered. Oesophageal achalasia is a rare cause of chronic cough in children, but it can mimic asthma symptoms presenting with stridor and acute respiratory insufficiency, and it may occur even in individuals with obesity.

DISCLOSURE

The authors declare no conflict of interest.

REFERENCES