The missing link

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Abstract

We present a case of a 28-year old woman who presented with bizarre wheezing breath sounds on expiration and dysphagia, with unexplained significant dilation of the esophagus mimicking achalasia finally leading to the diagnosis of a very small congenital tracheoesophageal fistula (TEF). Congenital TEF is usually detected shortly after birth and is typically accompanied by esophageal atresia. Congenital TEF without esophageal atresia (H-type fistula) can be missed in early life and diagnosis may be postponed until adulthood due to subtle symptoms. Diagnosis is usually based upon a combination of esophagoscopy, bronchoscopy, barium esophagography and CT-scan. The only clue can be the finding of a significant dilated aperistaltic esophagus, with subsequent more detailed CT reconstruction revealing a very tiny H-type TEF. It is important to raise the awareness of small H-type TEF as a possible cause of achalasia-like esophageal dilation in adulthood and of very unusual and bizarre wheezing breath sounds. (Acta gastroenterol. belg., **2018,** 81, **531-533**).

Key words : adult ; congenital ; tracheoesophageal fistula ; H-type.

Introduction

Most patients with congenital TEF are diagnosed shortly after birth or in early life due to frequent association with esophageal atresia which leads to life-threatening complications and warrants immediate surgery. (1) H-type TEF, where esophageal atresia is absent, is rare and diagnosis may be postponed until adulthood due to subtle symptoms and physician unfamiliarity (2). Until present, only 20 cases of H-type TEF diagnosis in adults have been described in the Anglo-Saxon literature. We report a case where finally the diagnosis of a congenital TEF was made in a healthy 28-year old woman.

Case report

A 28-year old woman presented at the outpatient clinic with a wheezing breath sound on expiration since childhood, and progressive hoarseness and intermittent dysphagia since 2 years. Medical history revealed appendectomy, frequent respiratory infections during childhood until the age of 10 years and viral meningitis at age 18. There is no history of smoking, alcohol or substance abuse. She uses no medication. Blood analysis, esophagoscopy with routine esophageal biopsies, lung function (pre- and post bronchodilatation), and routine CT-scan of the neck was normal. There were no anatomical abnormalities on barium esophagography, but a slightly atone aspect of the proximal esophagus

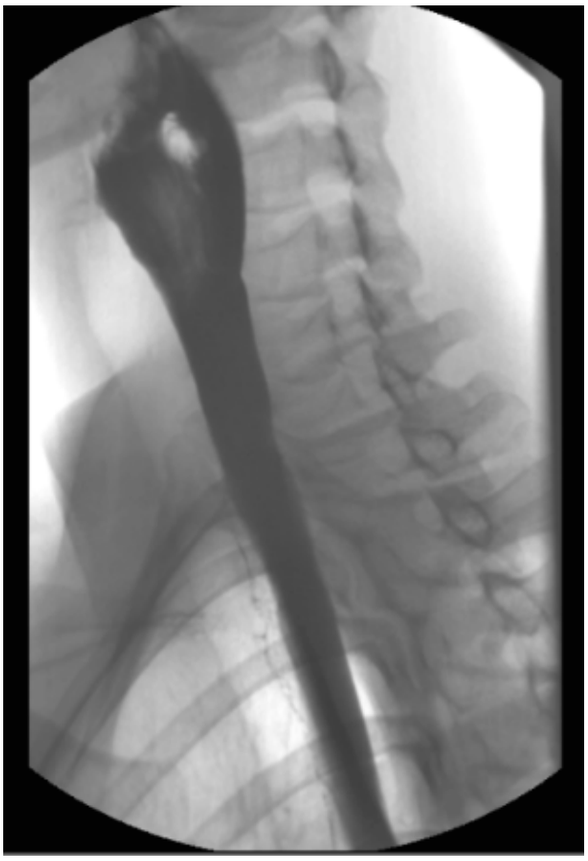

Fig. 1 — Barium esophagography : atone aspect of the proximal esophagus.

was noticed (Figure 1). Esophageal manometry was consistent with 100% aperistalsis in the esophageal body further confirming an atone esophagus, pressure in the lower esophageal sphincter was normal. High resolution CT scan (HRCT) of the thorax showed a very small esophageal diverticulum, an enlarged atone aspect of the esophagus and a subtle tracheoesophageal fistula (Figure 2). Finally, diagnosis of a congenital tracheoesophageal fistula with concurrent esophageal dysmotility was made. Due to subtle symptoms, absence of respiratory infections after age of 10 years, the high risk of persistent dysmotility after repair and preference of the patient

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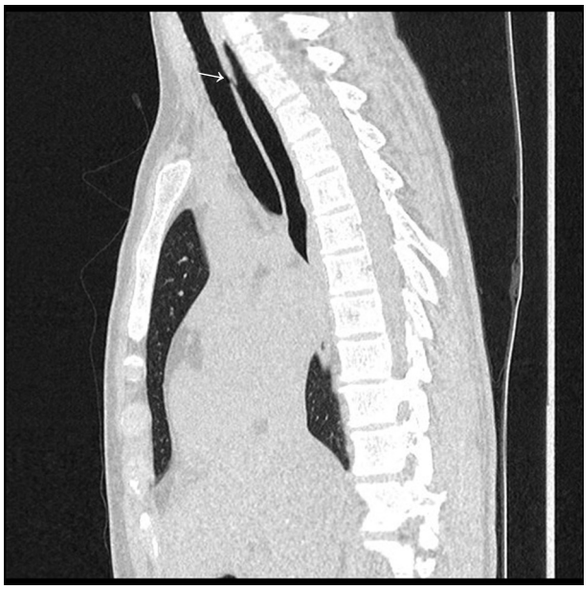

Fig. 2. — High resolution CT-scan of the thorax (HRCT) : subtle tracheoesophageal fistula (arrow).

we proposed conservative follow up. Until present, 6 months after initial diagnosis, our patient is doing fine.

Discussion

TEF was first reported as an incidental postmortem finding in 1929 by Negus. (3) Presentation of TEF in adults is predominantly associated with esophageal cancer. Less common, TEF is associated with trauma, prolonged intubation of the trachea and infections such as histoplasmosis, tuberculosis and actinomycosis. (2) Rare etiologies include Barrett's ulceration of the esophagus, ulceration of heterotopic gastric mucosa and TEF as an iatrogenic complication of endoscopic variceal sclerotherapy. Congenital TEF usually is associated with esophageal atresia and is typically diagnosed shortly after birth. Congenital TEF without esophageal atresia is called H-type TEF, due to the tracheal opening which is classically above the esophageal opening, giving the fistula the appearance of a slanted 'H'. (4) H-type TEF is very rare in adults. Until present, only 20 cases of congenital H-type TEF are described in Anglo-Saxon literature. The esophagus and trachea both develop from the primitive foregut. During the fourth and fifth week of embryologic development, the caudal part of the foregut (primitive pharynx) forms a ventral diverticulum that evolves into the trachea. The longitudinal tracheoesophageal fold fuses to form a septum that divides the foregut into a ventral laryngotracheal tube and a dorsal portion, the esophagus. The posterior deviation of the tracheoesophageal septum causes esophageal atresia and incomplete separation of the esophagus from the laryngotracheal tube resulting in a concurrent TEF. (7) Five main categories of esophageal atresia with our without tracheoesophageal fistula are described. (6)

Braimbridge and Keith (1966) classified congenital fistulas as Type 1 (2%): oesophageal diverticulum with a large ostium and a fistula at its tip. Type 2 (90%) : a short communication running directly from the oesophagus to the bronchus or trachea. Type 3 (3%): a fistulous tract connecting the oesophagus to a cyst in the lobe, which in turn communicates with the bronchus. Type 4 (5%) : a fistula that runs into a sequestrated segment or lobe. (5) The H-type fistula comprises only 4% of tracheoesophageal anomalies. (7) Due to minimal symptoms and physician unfamiliarity, diagnosis of H-type TEF may be postponed until adult life. Different theories are proposed to explain this often long silent interval until adulthood : 1) oblique course of the fistula which may close during swallowing, preventing aspiration 2) the presence of an occlusive membrane that covers the tract until adulthood and later ruptures 3) the presence of a proximal fold of esophageal mucosa that functions as a valve; after years of chronic inflammation this valve may become dysfunctional leading to the fistulous tract opening 4) spasm of the fistulous tract 5) gradual reduction in tone of the lower esophageal sphincter with age leading to increased reflux and hence soiling of the trachea. (2,5,8) When diagnosis of TEF is made, the proposed criteria for the definite diagnosis of a congenital etiology in adults are the following 1) absence of past or present surrounding inflammation or malignancy 2) complete recovery after resection 3) presence of normal mucosa and absence of adherent lymph nodes. (9)

Respiratory symptoms are usually more prevalent than gastrointestinal symptoms. A paroxysmal chronic cough, aggravated on swallowing liquids (called Ohno's sign) in combination with recurrent respiratory tract infections due to spillage of gastric contents in the respiratory tract, are pathognomonic. (2,10,11) Recurrent respiratory tract infections may, in the long run, lead to bronchiectasis and lung abscesses. (4) Less frequent symptoms may include blood-tinged sputum, retrosternal pain and halitosis. Sometimes, dysphagia, due to a dilated esophagus with aperistalsis, is the presenting symptom. (7) Disruption in utero of the normal development of the esophageal myenteric plexus causes disordered peristalsis and sometimes impaired lower esophageal sphincter function. (12) This explaines the aperistaltic, dilated esophagus with concurrent proximal esophageal diverticulum in our case. Associated developmental abnormalities are seen in 23% of cases of H-type TEF. A mnemonic, VACTERL (Vertebral Anorectal Cardiovascular Tracheo-Esophageal fistula Renal Limb) has been developed to help remember these associations. (13,14) Considering this variety of possible, often subtle and aspecific symptoms, close questioning of the patient is very important and can often trace symptoms back to childhood, raise suspicion for the diagnosis and guide further examination.

Diagnostic confirmation can be difficult. Barium esophagography in prone position is considered as the

gold standard for direct visualization of the fistula. At the same time, diagnosis of esophageal dilatation, achalasia and dysmotility, associated with the fistula, can be made. However, barium esophagography may not always provide definite diagnosis due the lower positioning of the esophageal opening relative to the tracheal opening which prohibits contrast flow from the esophagus into the trachea. High resolution CT-scan can be particularly useful since it may show the exact location of the fistula and assess possible damage of the lung parenchyma. (2,4,9,12) Chest X-ray may show a dilated esophagus due to associated aperistalsis. This 'achalasia-mimic' can be confirmed by esophageal manometry. Esophagoscopy and bronchoscopy are necessary to exclude alternative diagnosis but often fail to visualize the suspected fistula because it has a flap membrane which may cover and conceal its entrance. Moreover the location of the fistula in the upper third part and on the anterior wall of the esophagus, considered the 'black box' during esophagoscopy, makes diagnosis even more challenging. (12)

There are no official guidelines for the management of this very infrequent pathology. However, there seems to be no controversy in existing literature about the necessity for surgical management of congenital TEF. (2) Especially in presence of recurrent respiratory infections, postponing treatment may lead to chronic damage of the lung parenchyma with development of bronchiectasis and lung abcesses. Two different approaches are proposed. Thoracotomy is the most frequently used approach and is the only option when dealing with damaged lung which may need resection. However, a cervical approach along the border of the sternocleidomastoid muscle is adequate for most simple TEF repairs. Specifically, because most H-type TEFs occur in the upper half of the trachea, a cervical incision provides an excellent exposure for surgical repair. After dissection and division of the fistula, a muscle or pleural flap is recommended to prevent relapse. (2,8,15).

Chang-hua *et al.* propose endoscopic treatment of the congenital tracheoesophageal fistula with submucosal dissection (17).

Prognosis is good after surgery. However, esophageal dysmotility may persist even after the repair. In our case, due to only intermittent symptoms, absence of major complications, the high risk of persistent dysmotility after repair and preference of the patient, no surgery was performed (1,4).

Conclusion

TEF without esophageal atresia, called H-type TEF, is a very rare pathology that can present in adulthood with subtle respiratory and gastrointestinal symptoms and possible complications such as recurrent respiratory infections, bronchiectasis and lung abcesses. Importantly our case illustrates that congenital H-type TEF also should be considered as a potential cause of unexplained achalasia-like dilation of the esophagus , prompting to evaluating CT in more detail with reconstructions. Furthermore finding H-type TEF as the cause of a very bizarre wheezy noise in our patient, reminds us that not all that wheezes is asthma.

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