CASE REPORT

A delayed diagnosis of Mounier-Kuhn syndrome

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SUMMARY

Following a provisional diagnosis of asthma of several years' duration by his general practitioner, a 43-year-old otherwise healthy man who was a non-smoker was referred to a pulmonologist with worsening productive cough and exertional breathlessness. A thoracic CT scan revealed dilated airways (tracheal diameter 35 mm, left bronchial diameter 20 mm, right bronchial diameter 18 mm). Inflamed and easily collapsible airways were seen on bronchoscopy. The patient remained stable and was followed up with regular spirometry. A follow-up CT scan 7 years later showed tracheobronchomegaly (tracheal diameter 42 mm, left bronchial diameter 25 mm, right bronchial diameter 23 mm) with large cystic spaces consistent with Mounier-Kuhn syndrome. Repeat bronchoscopy showed a massively dilated trachea and generalised collapse on expiration with a dilated thin-walled bronchial tree. He was deemed ineligible for lung transplantation due to the extent of airway involvement making it difficult to anastomose donor lung to native tissue.

BACKGROUND

Mounier-Kuhn syndrome is often diagnosed late with few treatment options available. Sharing our local experience of this rare condition will help increase knowledge about it and make physicians aware of this chronic disease, thus ensuring early diagnosis and referral for lung transplantation.

CASE PRESENTATION

A 43-year-old man who was a lifelong non-smoker presented with symptoms of worsening cough and exertional breathlessness after he was diagnosed by his general practitioner with asthma of several years' duration. He was previously healthy and had no family history of note. Following failure to control his symptoms, he was referred to a pulmonologist.

INVESTIGATIONS

An initial thoracic CT scan revealed cystic bronchiectasis and bronchoscopy showed easily collapsible and inflamed airways. Sweat test, cystic fibrosis transmembrane conductance regulator gene testing, autoimmune screen and avian precipitins were negative. A follow-up CT scan 7 years later showed tracheobronchomegaly with large cystic spaces consistent with Mounier-Kuhn syndrome (figures 1 and 2). Repeat bronchoscopy showed a massively dilated trachea and generalised collapse on expiration with a dilated thin-walled bronchial tree (video 1).

Figure 1 Follow-up CT scan of this patient diagnosed with Mounier-Kuhn syndrome.

DIFFERENTIAL DIAGNOSIS

The clinical presentation may resemble a wide spectrum of cardiopulmonary diseases such as coronary artery insufficiency, congestive heart failure, congenital heart disease, severe chronic obstructive airways disease, bronchiectasis and pneumoconiosis.

TREATMENT

The patient has received several intravenous antibiotic courses for *Alcaligenes xylosoxidans*, methicillin-resistant *Staphylococcus aureus* (MRSA), *Achromobacter denitrificans* and *Pseudomonas aeruginosa*. He receives regular chest physiotherapy and is on continuous oxygen therapy, inhaled corticosteroids, bronchodilators and colomycin for *Pseudomonas* colonisation. He was intolerant to non-invasive ventilation.

OUTCOME AND FOLLOW-UP

Subsequently, the patient's pulmonary function has continued to deteriorate (forced expiratory volume

Figure 2 Follow-up CT scan of this patient diagnosed with Mounier-Kuhn syndrome.

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Video 1 Bronchoscopy of this patient diagnosed with Mounier-Kuhn Syndrome.

in 1 s (FEV₁ 1.5 L), forced vital capacity (FVC) 2.29 L, FEV₁/FVC 66%). He is followed up on a regular basis with repeat sputum cultures. Follow-up CT scan 3 years later showed worsening dilation of the airways (tracheal diameter, left bronchial diameter, right bronchial diameter).

DISCUSSION

Mounier-Kuhn syndrome is a rare condition characterised by marked tracheobronchial dilation and recurrent lower respiratory tract infections. Diagnosis is typically made in the third or fourth decade of life using CT and bronchoscopy¹ and is associated with substantial morbidity and mortality.² Diagnostic criteria on CT scan include a tracheal diameter of more than 30 mm, a right bronchial diameter of 20 mm and a left bronchial diameter of 18 mm.¹ This diagnosis should be considered in instances of recurrent pneumonia and mucous plugging.³ Therapy is mainly supportive. Lung transplantation provides no proven benefit with regard to risk of morbidity and death.

Literature reports have described a delayed onset of the disease typically during the third decade of life,^{1 3} as was the case in our patient. This prolonged evolution time suggests an interaction between genetic predisposition to the disease and acquired aetiology. Although 7 years might seem a short period of evolution in our patient, chronic inflammation especially due to recurrent chest infections might have accelerated the disease process.

Learning points

- ▶ Mounier-Kuhn syndrome is a rare condition which may be misdiagnosed as a more common pulmonary condition. We recommend that this condition should be considered in the differential diagnosis when a patient presents with recurrent respiratory tract infections.
- ▶ Limited treatment options are available. Increased knowledge of the condition may provide us with improved treatment options.
- ▶ Increased awareness of this condition as well as the availability of endoscopic imaging may prompt physicians to make an earlier diagnosis with the possibility of referral for transplantation at an earlier stage of the disease.

Competing interests None.

Patient consent Obtained.

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