ORIGINAL RESEARCH LETTER

Tracheobronchomegaly, cough and recurrent chest infection: Mounier-Kuhn syndrome

To the Editor:

A 49-year-old male ex-smoker was referred for recurrent chest infections requiring one course of antibiotics every winter, occurring over the last 20 years. Each episode is characterised by a productive cough with purulent sputum along with difficulty breathing, chest tightness and fatigue, but without haemoptysis. On some occasions, these symptoms were preceded by fever and rhinorrhoea. Each episode lasted ∼10 days and responded well to antibiotics. He had never been hospitalised for these infections, nor received a chest radiograph or sputum microbiology. He had been told that he developed episodes of pneumonia in his first year of life. Apart from these yearly infections, the patient was asymptomatic during the rest of the year.

The patient was born at term (normal delivery) and all his childhood vaccinations were administered. He had no history of recurrent sinus, gastrointestinal, urinary, skin, joint or central nervous system infections. His only significant past medical history was the presence of gastro-oesophageal reflux diseases (GORD), which was controlled by daily rabeprazole, 20 mg once daily. He was otherwise well without a history of dry eyes, dry mouth, joint pain, difficulty swallowing or muscle pains.

The patient lived at home with his wife and two children, and worked as a sales manager. He was an ex-smoker with a 5-pack-year history and stopped 20 years ago. He had drunk one glass of wine per day for ∼10 years. He had no pets at home. There was no family history of any congenital, lung or infectious diseases. The patient regularly visited the gym and performed a combination of high-intensity cardiovascular and resistance training.

There was no evidence of clubbing, jaundice, anaemia or cervical lymphadenopathy. On auscultation, his chest was clear with good bilateral air entry, and no evidence of wheeze or crackles. Heart sounds were normal. There were no rashes or evidence of peripheral stigmata of connective tissue disease.

Spirometry performed in clinic showed a forced expiratory volume in 1 s (FEV_1) of 4.43 L (122% predicted), forced vital capacity (FVC) 5.01 L (112% predicted), FEV_1/FVC ratio of 0.88 and a peak expiratory flow rate of 11.33 $L{\cdot}s^{-1}$ (122% predicted).

Full blood cell count, urea and electrolytes, liver function tests, C-reactive protein, complement, and immunoglobulin levels were all within normal ranges with no clinically significant abnormalities. B-cell and T-cell subsets were also normal.

A posterior–anterior chest radiograph showed evidence of tracheomegaly (figure 1a) with no other intrathoracic abnormalities. High-resolution computed tomography (CT) showed dilation of the trachea, mainstem bronchi, bronchial branches and borderline bronchiectasis. The trachea was noted to be dilated by 47 mm and 42 mm in the anteroposterior and transverse dimensions respectively (figure 1b). The right mainstem bronchus had a maximal diameter of 15 mm while the left mainstem bronchus had a maximal diameter of 16 mm (figure 1c and d, respectively). There were a few thin septations of the dilated trachea

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Mounier-Kuhn Syndrome (MKS) is a rare disease characterised by recurrent chest infections, and dilation of the trachea and main bronchi, most likely to due to atrophy of elastic fibres
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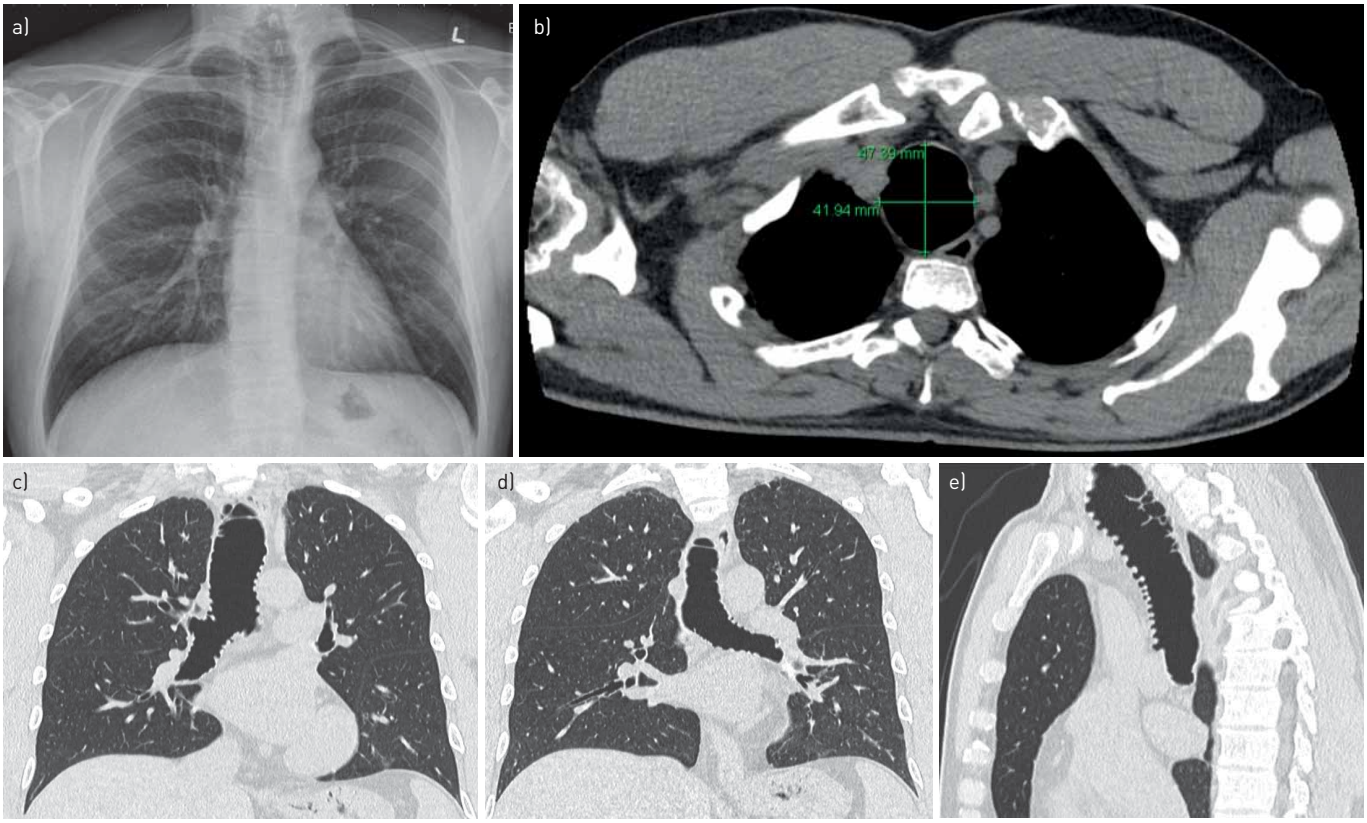

FIGURE 1 a) Chest radiograph (posterior–anterior) showing tracheomegaly. b) Computed tomography (CT) image of the trachea (axial view), largest diameter of trachea measuring 47mm×42 mm. CT image of the chest (coronal view) showing dilatation of c) right and d) left main bronchus. e) CT image of trachea (sagittal view) showing tracheal diverticula, septations and tracheomegaly.

with small diverticula present (figure 1e) along with a nodular appearance of the walls of the mainstem bronchi with small diverticula (figure 1c and d).

The clinical history of recurrent chest infections and combination of imaging findings was consistent with Mounier-Kuhn Syndrome (MKS). MKS is a rare disorder characterised by enlarged trachea and main bronchi. It was first described by Pierre-Louis Mounier-Kuhn in 1937 associated with recurrent chest infections [1] and anatomically described as tracheobronchomegaly (TBM) in 1962 [2]. This is a condition which is thought to occur because of atrophy of the elastic fibres [3] of the trachea and bronchi, leading to thinning of the smooth muscle, and ultimately causing the trachea to become flaccid, dilated and occasionally develop tracheobronchomalacia [4–6]. Weakness in the muscle layer causes diverticulum to form between the cartilaginous rings [1, 2, 7]. The diagnosis can be made on CT by measuring the diameter of the trachea, with some differences observed between sexes; in males, the trachea diameter is > 27 mm, whilst in females, measurements are slightly lower but exceed 23 mm [8, 9].

There has been some debate whether MKS is congenital or acquired. In a review of 128 cases over a 30-year period, the average age of diagnosis was 54 years with few symptoms during childhood [9]. However, in our case, the patient had been symptomatic since his mid-20s and undiagnosed. Chronic infection has been proposed as the cause of TBM, whilst others argue it can exist even without chronic infection [10–14]. Histological changes consistent with MKS can has been demonstrated in the absence of inflammation, suggesting recurrent infections are a consequence of the disease and not the primary cause [15–18]. A more functional classification based on age, pulmonary complications and evidence of elastosis been recently proposed [19].

It is thought that MKS could be inherited as an autosomal recessive trait as cases have been detected in siblings and cousins, but no gene has yet been identified [4, 8]. This condition is eight times more common in males [9], and has been associated with other connective tissue diseases such as Ehlers–Danlos syndrome [20] and cutis laxa in children [21], but only as single case reports.

A wide range of pulmonary and non-pulmonary clinical consequences related to MKS have been described [22]. The most common respiratory complications were bronchiectasis, tracheobronchomalacia and

emphysema, while the most common nonrespiratory comorbidity was GORD [23]. The presence of GORD in this patient may increase the risk of aspirations. In this case, our patient was asymptomatic for most of the year, except for short periods of infection, but there was no evidence of any significant bronchiectasis either clinically and radiologically. A possible reason for this could be the high peak flow rates and ventilatory capacity observed, which allows much easier clearance of respiratory secretions.

From a physiological perspective, flow is inversely proportional to radius to the fourth power. Hence, a doubling in the tracheal radius from 20 mm to 40 mm in our patient should result in a reduction in flow rates by 16 times. This would make clearing the airways more difficult. However, this was not demonstrated, and in fact, his flow rates were higher than normal predicted values, ~11 $L{\cdot}s^{-1}$.

The cross-sectional area of the patient's trachea was ~14 cm^2 based on an approximate radius of 2.1 cm. A normal patient would have an approximate cross-sectional area of 3.5 cm^2, based on a radius of 1.05 cm, which is half that of our patient. If the cross-sectional area has increased four-fold, then the flow rates would have to increase 16-fold, which would be required to maintain clearance of particles of the trachea. In our patient, the flow rate was 11 $L{\cdot}s^{-1}$, which is the double normal value and is likely close to the limit of being able to prevent the sequelae of the enlarged trachea. We speculate that flow rates would be a good index of prognosis for monitoring such patients.

Treatment of MKS is mainly supportive, which includes preventing and treating infections with vaccinations and antibiotics, along with clearing secretions with mucolytics and chest physiotherapy. In patients with tracheobronchomalacia, long-term continuous positive airway pressure [24], airway stenting [25, 26], surgical tracheoplasty [27] and laser treatment [28] have all been tried. Caution must be exercised as stenting can often lead to infection, obstruction and stent migration. Double lung transplantation has been performed in end-stage disease [29].

MKS is a rare disease characterised by recurrent chest infections and dilation of the trachea and main bronchi, most likely to due to atrophy of elastic fibres. The diagnosis should be suspected with a history of recurrent chest infections and radiographic evidence of tracheomegaly, especially in males. Treatment is aimed at reducing infections and improving airway clearance. In the patient described, it is speculated that he avoided many of the severe lung consequences of MKS because of exceptionally high flow rates helping to clear secretions.

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