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Beyond recurrent infections: a rare case of Mounier-Kuhn syndrome with tracheobronchial dilatation

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Abstract

Mounier Kuhn syndrome (MKS), also known as tracheobronchomegaly (TBM), is a very rare and chronic airway disease characterized by marked dilatation of the trachea and central bronchi. Currently, there are few epidemiological studies on MKS, and most data are derived from case reports, leading to a limited understanding of the disease by clinicians.

In this article, we present a 45-year-old male patient with MKS who had been symptomatic for a long time and had undergone various treatments due to a delay in diagnosis.

This case highlights the importance of considering Mounier-Kuhn syndrome in patients with recurrent or unexplained respiratory infections to ensure timely diagnosis and prevent unnecessary treatments.

Keywords Mounier Kuhn syndrome, Tracheobronchomegaly, Bronchiectasis, Recurrent infection

Introduction

Mounier Kuhn syndrome (MKS) is a rare clinical condition that is both late and misdiagnosed. It was first identified by Czyhlarz in an autopsy in 1897 but was first described by Pierluigi Mounier Kuhn in 1932 as a dilatation of the trachea and main bronchi with recurrent pulmonary infections [1, 2]. To date, 400 cases of Mounier Kuhn syndrome have been described [1]. There is no large, population-based study to determine the prevalence of MKS, but its prevalence is estimated to be 0.4–1.6% [3]. Risk factors for the disease include smoking and exposure to pollutants, which themselves cause chronic inflammation in the airways [4]. MKS is usually sporadic in adults; however, autosomal recessive familial cases and associations with disorders such as ataxia–telangiectasia, ankylosing spondylitis, Ehlers–Danlos syndrome, Marfan syndrome, Kenny–Caffey syndrome, Brachmann–de Lange syndrome, and cutis laxa (elastolysis) have been reported [5, 6].Several respiratory and systemic conditions—including cystic fibrosis, bronchiectasis, emphysema, chronic bronchitis, amyotrophic lateral sclerosis, sarcoidosis, giant cell arteritis, amyloidosis, rheumatoid arthritis, ankylosing spondylitis, Sjögren syndrome, and homocystinuria—have been reported in patients with tracheal or bronchial dilatation; however, these conditions are more often considered causes of

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secondary (acquired) dilatation rather than true associations with MKS [7–11].

It is important to note that tracheobronchomegaly may also develop secondary to acquired conditions, such as prolonged mechanical ventilation with excessive cuff pressure *(more than 30 cmH2O)*, radiotherapy for head and neck malignancies and fluoroquinolone complications [12–16]. These forms represent secondary tracheobronchomegaly rather than true congenital Mounier-Kuhn syndrome and should be considered in the differential diagnosis.

Despite increased awareness, MKS remains largely underdiagnosed because its clinical presentation often overlaps with more common chronic airway diseases such as chronic bronchitis and bronchiectasis [17]. Radiologic evaluation plays a pivotal role in identifying tracheobronchial dilatation and associated structural abnormalities that confirm the diagnosis [18]. We report a middle-aged man in whom MKS was diagnosed radiologically, with delayed clinical recognition of the condition.

Case presentation

A 45-year-old non-smoking male postal driver presented with recurrent respiratory infections, mild dyspnea, and productive cough without fever. He denied any relevant occupational exposures and had no significant family history. He is currently suffering from mild dyspnea, productive cough without fever. On brief examination, crackles are heard at the base of the left lung. On Chest x-ray (CXR) imaging, the diameter of the trachea is greater than the width of the vertebral column. High-resolution computed tomography (HRCT) of the lungs demonstrated marked dilatation of the central airways, with the tracheal, right main bronchial, and left main bronchial diameters measuring 36 mm, 26 mm, and 25 mm, respectively. In addition, cystic bronchiectasis with associated ground-glass opacities and centrilobular nodules was identified in the left lower lobe, findings consistent with bronchiolectasis and retained secretions (Fig. 1).

Pulmonary function testing revealed an obstructive ventilatory pattern, with an FEV1/FVC ratio of 66%, FEV1 of 62% predicted, and FVC of 77% predicted. Bronchoscopic examination demonstrated dilatation of the trachea and main bronchi with multiple diverticula. Bronchoalveolar lavage (BAL) analysis was negative for Mycobacterium tuberculosis.

Following the diagnosis of MKS, the patient was treated with antibiotics, bronchodilators, and mucolytics. Supportive care measures, including annual vaccination and avoidance of known allergens, were also recommended.

Discussion and conclusion

MKS, which is the dilation and enlargement of the trachea and main bronchi up to the fourth generation of the airway, is due to congenital weakness and atrophy of the elastic fibers and muscles of the tracheal wall and main bronchi [19–25].Weakness of the muscular layer is the cause of diverticula between the cartilage rings, especially the posterior wall of the trachea and main bronchi [26]. Airways distal to the fourth and fifth generations usually have normal diameters. The manifestations of the disease range from asymptomatic to respiratory failure due to recurrent pulmonary infections [26, 27].

MKS is more common in men, with an 8-fold higher incidence and is more common in the 3rd and 5th decades. Nonspecific symptoms include cough, purulent sputum, and dyspnea [5, 26] and cough is ineffective due to airway collapse [27].

MKS has 3 subtypes [22]:

1. Symmetrical and brief dilatation of the trachea and right and left main bronchi

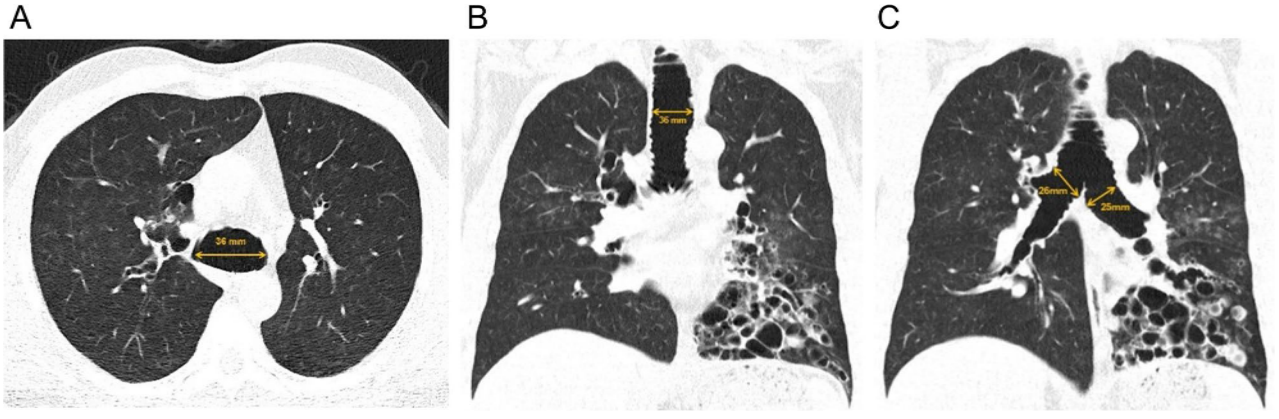


Fig. 1 HRCT images of the chest demonstrating classical findings of MKS. **A** Axial (transverse) section showing marked tracheal dilatation (36 mm). **B** Coronal reconstruction confirming tracheal enlargement. **C** Coronal view demonstrating enlargement of the main bronchi (right: 26 mm; left: 25 mm), with associated bronchiectatic changes.

2. Dilatation of the large airways with diverticula (most common type)
3. Saccular and diverticular structures extending to the distal bronchi

The diagnosis of MKS Tracheobronchomegaly is suggested by the appearance of an enlarged tracheal diameter on CXR, which is usually greater than the diameter of the spine [2, 28], but the gold standard for diagnosis is CT scan [29, 30].

Diagnostic criteria on lateral CXR are: tracheal diameter > 30 mm and diameters of the right and left main bronchi > 24 mm and 23 mm, respectively [31]. In addition, tracheal cross-sectional area can also confirm the diagnosis: > 371 mm^2 in men and > 299 mm^2 in women [5, 28].

Attention to respiratory rotation is important in the diagnosis of MKS on CT scan because the difference in bronchial diameters during inspiration and expiration can be effective in the differential diagnosis. Bronchoscopy plays a role in the diagnosis of tracheal and main bronchial dilation during inspiration and airway collapse during expiration and cough due to tracheomalacia [32]. It should be remembered that the size of the tracheal diameter depends only on gender and not on weight or height [33].

Differentiating MKS from bronchiectasis is important because bronchial ectasia is associated with MKS. Bronchiectasis affects the segmental and subsegmental bronchi, whereas bronchiectasis with MKS is seen only in the trachea and main bronchi.

Physiologically, airflow is inversely proportional to the fourth power of the radius of the airways. If the radius of the trachea is doubled, the flow velocity decreases 16-fold. Reduced airflow velocity leads to dysfunction of the mucociliary system, increased difficulty in clearing the airways, and ultimately airway obstruction with mucus [34].

Diverticula, which are the result of weakness in the muscular layer of the airway wall, are mostly located in the posterolateral wall [4, 12]. Diverticula sometimes act as reservoirs for secretions and increase susceptibility to recurrent pulmonary infections and bronchiectasis [35].

Pulmonary function tests (PFTs) typically demonstrate an obstructive ventilatory defect, characterized by increased residual volume (RV) and total lung capacity (TLC), along with reduced diffusing capacity for carbon monoxide (DLCO). In advanced disease, a mixed obstructive and restrictive pattern may be observed [20].

The following cases should be considered in the differential diagnosis [36]: William Campbell with congenital bronchiectasis cysts due to a lack of cartilage in the airways of the 4th-6th generation onwards, in which the trachea and main bronchi are normal [2].

Laryngocele, pharyngeal diverticula, and Zenker diverticulum represent mucosal protrusions of the larynx or pharynx and are not associated with tracheal diverticula [37, 38].

Bullous emphysema resulting from damage to the alveolar wall [39, 40] and apical lung herniation as protrusion of the lung from the thoracic wall [41], in which the trachea and main bronchi are normal in both cases.

MKS presents challenges in the anesthesia and intubation process, including a dilated airway with a weak and fragile wall and diverticula, which can cause ineffective coughing and retention of secretions, which increases susceptibility to infection. An indirect trachea can make intubation difficult and ultimately increase the risk of postoperative tracheal collapse [41]. It should be noted that sometimes the first sign of MKS is a leak of the endotracheal tube cuff during intubation.

In the treatment of MKS, the following should be observed:

1- Quitting smoking and reducing exposure to allergens, irritants, and occupational pollutants
2- Annual and mandatory influenza and pneumococcal vaccination
3- In cases of exacerbation, prescribing antibiotics, bronchodilators, chest physiotherapy, and postural drainage
4- Finally, in patients with resistant and recurrent symptoms, surgical stenting, tracheobronchoplasty, and transplantation [4].

Our patient remained undiagnosed for years despite recurrent respiratory infections. This case emphasizes the need for increased awareness of MKS and the role of radiologic and bronchoscopic evaluation in achieving timely diagnosis and improved outcomes.

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Authors' contributions
Mohsen Sadeghi: conceptualization, data curation, methodology, supervision, writing – review and editing. Fatemeh Sadat Hosseini Khajouei: data curation, methodology, writing – original draft. Nilsa Dourandish: validation, writing – review and editing. Amir Morteza Soleimani: Investigation, methodology, project administration, supervision, writing – original draft, writing – review and editing.

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Data availability
This published article contains all the data generated or analyzed during the present study.

Declarations

Consent for publication

Written informed consent was obtained from the patient for the publication of this case report and any accompanying images.

Competing interests

The authors declare no competing interests.

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