Received: 2019.07.05
Accepted: 2019.11.26
Available online: 2020.01.24
Published: 2020.02.09

Recurrent Pneumonias in a Previously Healthy and Immunocompetent Young Adult: A Case Report Mounier-Kuhn Syndrome

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Conflict of interest: None declared

Patient: Male, 45-year-old
Final Diagnosis: Mounier-Kuhn syndrome
Symptoms: Heart failure • pneumonia
Medication: —
Clinical Procedure: —
Specialty: Pulmonology

Objective: Rare disease
Background: Mounier-Kuhn syndrome is a rare clinical condition characterized by marked tracheal and bronchial dilatation. It is commonly associated with multiple respiratory infections due to the inability to mobilize secretions, chronic cough, and decline in lung capacity. Although it is a rare disease, the number of cases reported in the literature has increased, and physicians should be aware of its existence to be able to recognize and diagnose it. Suspicion should arise regarding young adults who develop recurrent airway infections when immunosuppression has been ruled out.

Case Report: We present the case of a young adult, previously healthy and immunocompetent, who was diagnosed with Mounier-Kuhn syndrome after 5 years of multiple respiratory infections and multiple respiratory failures, who received intensive care management with multiple complications and difficulties in achieving adequate ventilation. Due to the risk of respiratory failure and major complications, its management was challenging. We were able to treat this patient and prevent complications with a multidisciplinary approach with a strong emphasis on constant and intense pulmonary therapy.

Conclusions: Mounier-Kuhn syndrome is rare but does occur, and physicians need to have a high index of suspicion to diagnose it. Its management must be done by a multidisciplinary team, and respiratory therapy should be constantly available at bedside.

MeSH Keywords: Case Reports • Tracheal Diseases • Tracheobronchomegaly

Full-text PDF: <https://www.amjcaserep.com/abstract/index/idArt/918535>

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Background

Mounier-Kuhn syndrome, also known as tracheobronchomegaly, is a rare disease, first described in an autopsy in 1897 by Czyhlarz. However, it was not until 1932 that Dr. Mounier-Kuhn described the syndrome with particular clinical, radiological, and endoscopic characteristics [1,2]. A prevalence rate of 0.4–1.6% has been described in several series of cases, with a greater incidence in males, with a sex ratio of 8: 1. Due to its low incidence, it is usually not included in differential diagnoses, and it is often undiagnosed. However, there has been an increase in the number of case reports, which suggests that the prevalence is higher and it is a sub-diagnosed entity. To date, around 300 cases have been reported worldwide [3,4].

Its etiology is unknown and no gene mutation has been identified, although some cases have been described. Its histology, described in 1962 by Katz, shows absence of elastic fibers, atrophy of longitudinal muscles, and weakening of the smooth muscular layer, predisposing to dilatation of the cartilaginous and membranous area of the trachea and bronchi, generating mucous herniation and formation of secular diverticula between the cartilaginous rings [1,3]. There is also an increase in distensibility of central airway walls during inspiration, favoring collapsibility during expiration, especially during forced expiration and coughing. Concomitantly, mucociliary clearance decreases, which favors the accumulation of secretions and respiratory infections. All these pathophysiological changes lead to bronchiectasis and pulmonary capacity deterioration [2,3]. Pulmonary function tests often show increased dead space and tidal volume, with obstructive patterns and high total lung capacity. In advanced phases of the disease, a restrictive pattern can be seen.

Cases have been reported as young as 18 months up to 95 years. However, its incidence is highest in people in their 40s [5]. Some authors have described an association with smoking cigarettes, but this has not been corroborated [3,6].

Its clinical presentation can vary from an asymptomatic to chronic dry or moist cough, hemoptysis, progressive dyspnea, repeated respiratory infections, pneumonia with therapeutic failure, ventilatory failure, and even death [4,7]. Physical examination can evidence digital clubbing, rales and/or rhonchi on pulmonary auscultation, tachycardia, tachypnea, fever, and ventilatory failure [4].

There are several predisposing conditions, including pulmonary fibrosis, prolonged mechanical ventilation (especially in preterm infants), and neck radiotherapy [3]. It has been associated with various hereditary syndromes such as Ehlers-Danlos, Marfan, laxa cutis, Kenny-Cafey, Bruton's disease, ankylosing spondylitis, rheumatoid arthritis, non-small cell

carcinoma, ovarian cancer, T-cell lymphoma, and esophageal carcinoma [3,4,6].

The clinical suspicion must be confirmed with diagnostic images. There are radiographic findings on chest x-ray, where the size of the trachea exceeds that of the vertebral bodies. However, a chest computed tomography (CT) scan is the criterion standard. So far, there are no diagnostic criteria according to the population and anthropometric characteristics; however, it is accepted that there should be a transverse tracheal diameter greater than 3 cm, right bronchial diameter greater than 2.4 cm, and left bronchial diameter greater than 2.3 cm [8]. Magnetic resonance imaging (MRI) does not seem to provide additional information, and some authors recommend that it should not be ordered [4,9].

Asymptomatic patients do not require treatment; however, in symptomatic patients, management with mucolytic, physical, and respiratory therapy is recommended. It is important to improve bronchial hygiene with postural drainage to counteract the decrease in mucociliary movement, and antibiotics should be used in case of infection. Vaccination against pneumococcus and influenza has been suggested, resulting in the prevention of exacerbations [7]. Bronchial plasty with silicone stent has been described and is associated with improvement compared to medical management alone; however, the availability of an appropriate stent can be a limiting factor for some institutions [4,7]. In more severe cases, lung transplantation has been considered, but unfavorable results have been reported [7,10,11]. The life expectancy of these patients has not been documented. Possible complications include obstructive pulmonary diseases, pulmonary fibrosis, pneumothorax, repeated respiratory infections, and chronic respiratory failure [3,7].

Here, we present the case of a young adult diagnosed after 5 years of symptoms, who presented with multiple respiratory infections and received intensive care management, with multiple complications and difficulties in achieving adequate ventilation.

Case Report

A 44-year-old man, previously healthy, with no family history of relevant diseases, was referred to the outpatient pulmonology service in 2015. He denied tobacco or alcohol consumption and had no occupational exposure to biohazards. His chief complaint was 5 years of multiple community-acquired pneumonias with concomitant requirement of in-hospital management on the general ward and intensive care units (UCI), with more than 9 hospitalizations in the past 5 years. His symptoms began 5 years ago with a common cold that lasted over 1 month. Subsequently, he had several colds that evolved to severe cases of pneumonia and he received multiple antibiotic

Figure 1. Posterior anterior chest radiography. Increased lung volume with cystic images in the lower two-thirds of the lungs, some with air-fluid levels due to infected bronchiectasis. Note the increased tracheal diameter.

regimens with treatment failure. Primary care physicians had ruled out immunodeficiency, tuberculosis, HIV, and even cystic fibrosis, without identifying the cause of his multiple pulmonary infections. Then, he began to have decreased cardiac functional capacity and required supplemental oxygen by nasal cannula. He had multiple intensive care unit admissions for pulmonary infections and cardiopulmonary complications.

During a medical appointment with a pulmonologist, he received an echocardiogram with evidence of severe pulmonary artery systolic pressure (PSAP) of 64 mmHg with preserved left ventricular ejection fraction (LVEF), arterial gases with severe hypercapnia, chest x-ray with bilateral apical interstitial infiltrates with multiple cystic areas, and chest CT with multiple bilateral basal cystic areas (Figures 1, 2).

He was diagnosed with advanced lung disease with permanent oxygen requirement, severe pulmonary hypertension, NYHA IV (New York Heart Association) functional class. Due to the severe lung impairment, he was referred to the emergency department to complete studies and evaluate the possibility of a 2-lung transplant. During the hospitalization, a CT scan was performed, showing a mega-trachea with a 3.48 cm diameter, tracheal diverticula, and cysts in the middle and basal segments, with precapillary pulmonary hypertension (Figures 2, 3). Bronchoscopy showed a mega-trachea, multiple tracheal diverticula, alteration of the bronchial anatomy with loss of segmental divisions, and multiple caverns filled with pus (Figure 4).

Figure 2. High-resolution chest CT scan. Cystic and cylindrical bronchiectasis in the middle third of the lungs, with tracheomegaly [23x35 mm].

Figure 3. 3D MIP lung and airway reconstruction of chest scans. Diffuse tracheobronchial dilatation with predominantly basal bronchiectasis.

The diagnosis of Mounier-Kuhn syndrome was made during this hospitalization in 2015 based on the finding of a mega-trachea with a diameter of 3.48 cm. The lung transplant was not possible due to the trachea diameter. The patient's condition was explained thoroughly to him and his family, as well

Figure 4. Bronchoscopy showing the trachea with a marked increase in diameter, multiple diverticula in the mucosa, and abundant purulent secretions in the distal trachea.

its life-threatening nature, and the symptomatic and preventive management that must be established to prevent exacerbations was also explained. He was discharged on salbutamol, mucolytic, and respiratory therapy once daily.

Since his first visit to the pulmonologist in 2015, he has been admitted to the emergency department 18 times, all of them due to exacerbation of respiratory symptoms associated with resistant microbial infections and dyspnea. He has received multiple antibiotic regimens accompanied by major respiratory therapy and it has been possible to avoid the requirement of orotracheal intubation by managing respiratory failures with non-invasive mechanical ventilation.

Discussion

Mounier-Kuhn syndrome is a rare disease characterized by marked tracheal-bronchial dilatation [1,2]. It is commonly associated with multiple respiratory infections due to the inability to mobilize secretions, chronic cough, and progressive decline in lung capacity [1,2]. The patient in this case report is

an example of a young adult with recurrent respiratory infections that began at his mid-40s without any associated risk factors. In clinical practice, and especially in the emergency department, it is very important that when a patient arrives with these symptoms the physicians try to rule out immunodeficiency, which is the primary cause of recurrent respiratory infections [9]. Once these conditions are ruled out, the physician should focus on possible anatomic conditions that may predispose to infections, and it at this point that these types of anatomic disturbances, like Mounier-Kuhn syndrome, should be considered. The first approach may be as simple as a chest x-ray and, as mentioned before, if the size of the trachea exceeds that of the vertebral bodies, the possibility of an anatomic problem with the trachea may be suspected and further imaging such as a CT scan can be ordered.

Management can be challenging due to the multiple complications that may arise, which is why a multidisciplinary team of emergency physicians, pulmonologists, anesthesiologists, and intensive care physicians should have sufficient knowledge and be prepared to manage these patients.

Our patient's age at diagnosis was similar to that reported by other authors, as most cases are diagnosed at ages 40–50 years [12,13]. Although most reported cases have chronic cough and the vast majority are asymptomatic, there are cases, like our patient, in which lung capacity is severely impaired [12,13].

Conclusions

Mounier-Kuhn syndrome is an uncommon and underdiagnosed entity. In the case presented here, the description of recurrent respiratory infections in a young immunocompetent patient without risk factors for pulmonary or infectious pathology should make us aware of possible anatomic causes of these symptoms. Physicians must be aware of the existence of this disease in order to diagnose it. Finally, use of strong and constant respiratory therapy allowed us to successfully to manage this patient.

References:

1. Jaiswal AK, Munjal S, Singla R et al: A 46-year-old man with tracheomegaly, tracheal diverticulosis, and bronchiectasis: Mounier-Kuhn syndrome. *Lung India*, 2012; 29(2): 176–78
2. Celik B, Bilgin S, Yuksel C: Mounier-Kuhn syndrome. *Tex Heart Inst J*, 2011; 38(2): 194–96
3. Katz I, Levine M, Herman P: Tracheobronchiomegaly. The Mounier-Kuhn syndrome. *Am J Roentgenol Radium Ther Nucl Med*, 1962; 88: 1084–94
4. Krustins E, Kravale Z, Buls A: Mounier-Kuhn syndrome or congenital tracheobronchomegaly: A literature review. *Respir Med*, 2013; 107(12): 1822–28
5. Simkins A, Maiti A, Cherian SV et al: Mounier-Kuhn syndrome. *Postgrad Med J*, 2017; 93(1104): 642
6. Severiche-Bueno D, Severiche-Hernández D, Severiche-Bueno D.F., Vragas M.T.: [Mounier Kuhn syndrome in a 78-year-old patient with pulmonary fibrosis.] *Acta Médica Colombiana*, 2017; 42(3): 198–201 [in Spanish]
7. Akgedik R, Karamanli H, Kizilirmak D et al: Mounier-Kuhn syndrome (tracheobronchomegaly): An analysis of eleven cases. *Clin Respir J*, 2018; 12(3): 885–89
8. Dunne MG, Reiner B: CT features of tracheobronchomegaly. *J Comput Assist Tomogr*, 1988; 12(3): 388–91
9. Kachhawa S, Meena ML, Jindal G, Jain B: Case report: Mounier-Kuhn syndrome. *Indian J Radiol Imaging*, 2008; 18(4): 316–18

10. Menon B, Aggarwal B, Iqbal A: Mounier-Kuhn syndrome: Report of 8 cases of tracheobronchomegaly with associated complications. *South Med J*, 2008; 101[1]: 83–87
11. Odell DD, Shah A, Gangadharan SP et al: Airway stenting and tracheobronchoplasty improve respiratory symptoms in Mounier-Kuhn syndrome. *Chest*, 2011; 140(4): 867–73
12. Fortuna FP, Irion K, Wink C, Boemo JL: Mounier-Kuhn syndrome. *J Bras Pneumol*, 2006; 32(2): 180–83
13. Ip JJK, Hui PKT, Lam SHY et al: Mounier-Kuhn syndrome: An unusual underlying cause for chronic coughs and recurrent pneumonias. *Hong Kong Med J*, 2013; 19(4): 4–5