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# A 58-year-old man with progressive dyspnea suffering from Munier-Kuhn syndrome.

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## Abstract

A 58-year-old man who was a former smoker and had a 20-pack-year smoking history presented to the hospital with worsening shortness of breath. Pulmonary function tests revealed mixed obstructive-restrictive physiology. Forced vital capacity (FVC) was predicted at 68%, forced expiratory volume in 1 second (FEV1) at 60% was predicted, FEV1/FVC at 67% was predicted, and Carbon monoxide (DLCO) diffusion capacity is expected to be 70%.

Chest images show increased interstitial density around the hilum. There is massive tracheobronchiomegaly with a tortuous appearance in the airways. There is also a small degree of parenchymal scarring and fibrosis-like reticular perihilar interstitial opacity. CT images show traction bronchiectasis is a finding in the setting of fibrotic lung disease. When lung tissue becomes inelastic due to the formation of dense fibrosis, the airways appear blocked and dilated. Although this patient has dilated airways, the bronchiectasis is greater than the small amount of parenchymal fibrosis seen. Therefore, this process is not caused by parenchymal fibrosis, but rather by inherent pathology of the airway wall. The most frequently reported symptoms of Mounier-Kuhn syndrome are severe cough, difficulty breathing, and recurrent respiratory infections. Patients may complain of chest pain and hemoptysis, but systemic symptoms are rare and require immediate investigation for other diseases. Pulmonary function tests often reveal obstructive pulmonary disease, but in advanced stages there may be limited parenchymal changes, resulting in a mixed appearance, as in a patient's case. Because airway changes are considered irreversible, treatment is aimed at managing symptoms and reducing long-term complications. Recommended lifestyle modifications include quitting smoking and avoiding irritants/particulates. Concurrent COPD or asthma is addressed to limit further morbidity. The infection is treated aggressively, usually requiring broad-spectrum

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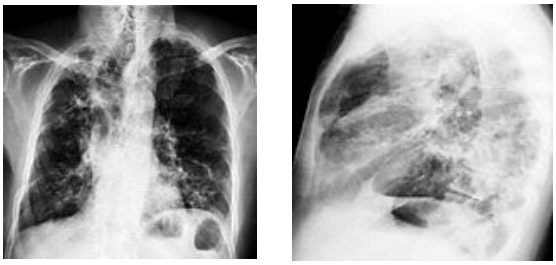
antibiotics to treat atypical infections. The use of expectorants and chest physiotherapy is considered helpful in preventing recurrent infections. Tracheal stenting may be considered if there are specific airways prone to collapse, but the spread of airway involvement limits its potential utility.

**Key word:** Dyspnea, Chest X-Ray, Chest CT, Munier-Kuhn syndrome

## 1. History and Radiogram

A 63-year-old man who was a former smoker and had a 20-pack-year smoking history presented to the hospital with worsening shortness of breath. Pulmonary function tests revealed mixed obstructive-restrictive physiology. Forced vital capacity (FVC) was predicted at 68%, forced expiratory volume in 1 second (FEV1) at 60% was predicted, FEV1/FVC at 67% was predicted, and Carbon monoxide (DLCO) diffusion capacity is expected to be 70%.

A chest radiograph was taken, showing postero-anterior and left lateral projections.



**(A) P-A** **(B) Left lateral**

Figure 1. Chest X-Ray Images

Figure 1 Chest images show increased interstitial density around the hilum. There is massive tracheobronchiomegaly with a tortuous appearance in the airways. There is also a small degree of parenchymal scarring and fibrosis-like reticular perihilar interstitial opacity. A noncontrast chest CT was performed to better evaluate the abnormalities seen on the chest radiograph.

Axial, sagittal, and coronal images are shown.

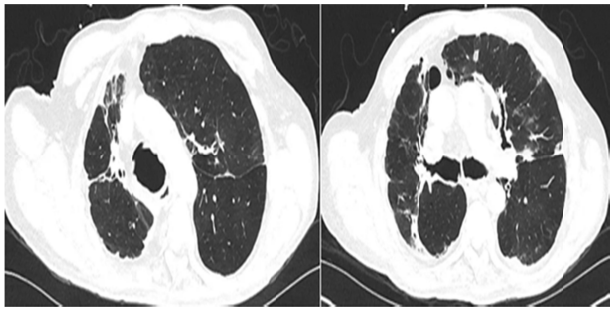


Figure 2. Axial scan images of chest CT

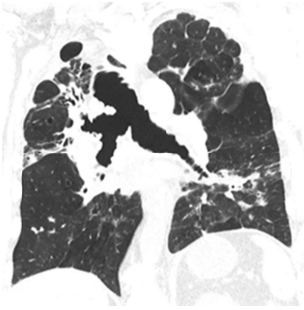


Figure 3. Coronal scan image of chest CT

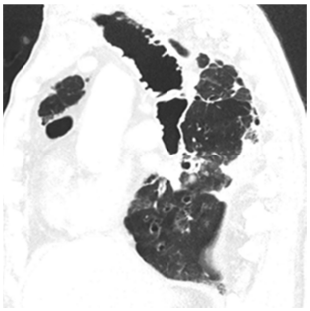


Figure 4. Sagittal scan image of chest CT

Traction bronchiectasis is a finding in the setting of fibrotic lung disease. When lung tissue becomes inelastic due to the formation of dense fibrosis, the airways appear blocked and dilated. Although this patient has dilated airways, the bronchiectasis is greater than the small amount of parenchymal fibrosis seen. Therefore, this process is not caused by parenchymal fibrosis, but rather by inherent pathology of the airway wall.

## 2. Finding

### Radiograph:

tracheobronchial hypertrophy

Thickening of bronchial walls

Interstitial opacities and atelectasis

### CT:

Organomegaly (33 mm in diameter)

Main bronchi dilatation (diameter 25 mm)

Bronchiectasis/bronchiolectasis

Tracheal/bronchial diverticula

Parenchymal scarring and volume loss

## 3. Differential Diagnosis

Cystic fibrosis

Mounier-Kuhn syndrome

Cutis laxa, Marfan syndrome, Ehlers-Danlos syndrome

COPD, bronchitis

Pneumoconioses

## 4. Diagnosis: Mounier-Kuhn syndrome

Munier-Kuhn syndrome, also known as tracheobronchiomegaly, is a rare medical condition characterized by enlargement of the trachea and main bronchi. This causes the airway walls to weaken and stretch. The disease was first described by Pierre Mounier-Kuhn in 1932. The main features of Munier-Kuhn syndrome include[(1)]:

**Bronchiectasis:** The trachea (trachea) and bronchi (airways leading to the lungs) become abnormally widened or enlarged.

**Weakened airway walls:** The walls of the trachea and bronchi become structurally weakened, resulting in reduced elasticity and muscle tone.

**Respiratory Symptoms:** People with Munier-Kuhn syndrome may experience a variety of respiratory symptoms, including chronic cough, excessive mucus production, recurrent respiratory infections, and difficulty breathing.

**Increased risk of complications:** Weakened airway walls increase the risk of complications such as recurrent pneumonia and bronchiectasis.

The exact cause of Munier-Kuhn syndrome is not well known. In some cases, there may be genetic factors, and in other cases, it may be caused by acquired factors. Smoking is considered a potential contributing factor. Diagnosis usually involves imaging tests such as a chest x-ray, computed tomography(CT) scan,

or bronchoscopy. Pulmonary function tests may also be done to evaluate lung function. Management and treatment of Munier-Kuhn syndrome focuses on resolving symptoms and preventing complications. Treatment may include medications to manage respiratory symptoms, airway clearance techniques, and in severe cases, surgery. Lung infections must be treated immediately to prevent further damage. Because Munier-Kuhn syndrome is a rare disease, comprehensive treatment often requires a multidisciplinary approach involving pulmonologists and respiratory therapists. Regular monitoring and individualized treatment plans are essential to manage the condition and improve the quality of life for affected individuals.

# 5. Discussion

## Mounier-Kuhn syndrome

### (1) Pathophysiology

Munier-Kuhn syndrome (MKS) is a disease primarily characterized by tracheal and bronchial wall enlargement. The exact cause is unknown, but it is likely to be due to genetic and/or epigenetic factors, hence the name congenital tracheobronchiomegaly. Pathological examination reveals that the characteristic airway dilatation may be preceded by atrophy of smooth muscle and elastic fibers within the larger airway walls. During inspiration, the airways remain open, but relaxed tracheal and bronchial walls may collapse during expiration. This can lead to obstructive pulmonary physiology, and patients often suffer from COPD in its early stages and are treated for it. Airway relaxation reduces particle removal effectiveness and increases the risk of pulmonary infection, especially for atypical bacteria in the lower respiratory tract. Recurrent infections can lead to bronchiectatic changes in small airways with parenchymal fibrosis, as seen in a patient.

### (2) Epidemiology

Approximately 300 cases have been described in the literature, with a ratio of affected males to females of 8:1. The average age of disease onset is the mid-50s, and the proportion of people with Mounier-Kuhn syndrome (MKS) peaks in their early 60s, as do patients. There are equal differences between smokers and non-smokers and may have some association with a predisposition to connective tissue disorders[(2)].

### (3) Clinical presentation

The most frequently reported symptoms are severe cough, difficulty breathing, and recurrent respiratory infections. Patients may complain of chest pain and hemoptysis, but systemic symptoms are rare and require immediate investigation for other diseases. Pulmonary function tests often reveal obstructive pulmonary disease, but in advanced stages there may be limited parenchymal changes, resulting in a mixed appearance, as in a patient's case.

### (4) Imaging Features

Imaging is often sought in patients with progressive respiratory distress or recurrent infections, and patients typically have a chest x-ray when symptoms appear. Patients may have symptoms that precede chest x-ray findings, but common findings are

tracheal enlargement and bilateral main bronchiectasis. Bronchiectasis may be present with reticular interstitial opacities and volume reduction, indicating post-inflammatory fibrotic changes. High-resolution CT can be diagnostic in the right clinical setting. It can be diagnosed when the tracheal diameter is more than 3 cm and the main bronchial tube diameter is 2 cm to 2.5 cm. Expiratory imaging may show collapse of the larger airways (i.e., crescentic trachea) and air trapping. Bronchiectasis is a hallmark and may present as clustered cystic structures in the subpleural space and may be confused with cystic lung disease or honeycombing. Patients with extensive bronchiectasis may be at increased risk for bronchopleural fistulas and recurrent pneumothorax, which should be considered in patients with a history of MKS and new chest pain. As in this patient, predominant fibrotic changes may be present around the hilum, and there may be varying degrees of volume loss, most likely a sequela of recurrent infection[3].

## (5) Differential Diagnosis

Although not as severe as MKS, several other congenital diseases can cause tracheobronchiomegaly. Cystic fibrosis can cause similar pulmonary pathology, but patients are younger, and the airways are typically affected, inflamed, and have extrapulmonary symptoms. Other congenital connective tissue disorders that cause similar airway changes include flaccid dermatosis, Ehlers-Danlos syndrome, Marfan syndrome, and ataxia telangiectasia. COPD causes similar symptoms and can cause acquired tracheomalacia. End-stage fibrotic lung disease can cause irreversible traction bronchiectasis and potentially tracheal dilatation. Moreover, prolonged mechanical ventilation can cause similar relaxation and airway dilatation[4].

## (6) Treatment

Because airway changes are considered irreversible, treatment is aimed at managing symptoms and reducing long-term complications. Recommended lifestyle modifications include quitting smoking and avoiding irritants/particulates. Concurrent COPD or asthma is addressed to limit further morbidity. The infection is treated aggressively, usually requiring broad-spectrum antibiotics to treat atypical infections. The use of expectorants and chest physiotherapy is considered helpful in preventing recurrent infections. Tracheal stenting may be considered if there are specific airways prone to collapse, but the spread of airway involvement limits its potential utility.

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