A Case of Mistaken Identity: Mounier-Kuhn Syndrome Masquerading as COPD

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INTRODUCTION

Mounier-Kuhn syndrome (MKS), or tracheobronchomegaly, is a rare congenital disorder characterized by dilatation of the trachea and main bronchi due to atrophy of smooth muscle and elastic fibers. The structural abnormality results in ineffective mucociliary clearance, predisposing patients to recurrent respiratory infections, progressive bronchiectasis, and chronic airflow limitation. Although well described in the literature, MKS remains underrecognized in clinical practice and is frequently misdiagnosed as asthma, bronchitis, or COPD. Non-smokers with chronic cough or unexplained bronchiectasis are particularly vulnerable to delayed or missed diagnosis, as clinicians may not consider congenital airway disease in the differential.

IMAGING

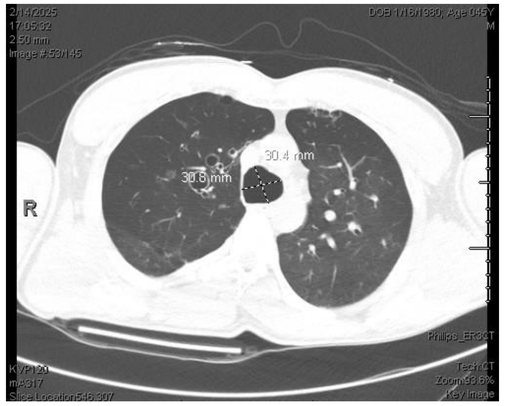

FIGURE 1: Axial CT showing tracheal enlargement

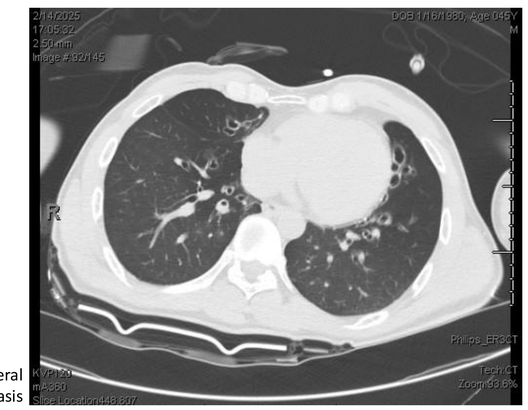

FIGURE 2: Axial CT showing bilateral bronchiectasis

CASE DESCRIPTION

A 45-year-old man with a history of “COPD” presented as a trauma alert after a heavy machine fell on him. On arrival to the emergency department, he was hemodynamically stable, afebrile, and maintained oxygen saturations of 95% on room air. Trauma evaluation revealed a T11 burst fracture managed with a TLSO brace and a right shoulder fracture dislocation treated with closed reduction.

Incidentally, chest CT demonstrated tracheobronchomegaly with a transverse tracheal diameter of 34 mm and bilateral bronchiectasis changes. Review of a prior high-resolution CT from 2017 revealed identical findings, confirming that these abnormalities had been present for years. Despite this, the patient had carried a diagnosis of COPD without ever being evaluated for congenital airway disease.

The patient reported lifelong daily productive cough, frequent respiratory infections, and progressive exertional dyspnea, which began in childhood and contributed to frequent missed school days. The patient has never smoked and did not report other risk factors for obstructive lung disease. Based on imaging and history, the diagnosis of Mounier-Kuhn syndrome with associated bronchiectasis was made.

The patient was started on an airway clearance regimen that included nebulized bronchodilators, an Acapella valve, and structured pulmonary hygiene education. Case management arranged for home nebulizer equipment, and plans were made for outpatient follow-up. Additional workup for potential secondary contributors to bronchiectasis, including immunologic and rheumatologic evaluation, was also initiated and came back negative.

DISCUSSION

This case underscores the diagnostic challenge of Mounier-Kuhn syndrome, which is often masked by overlapping symptoms of more common conditions, such as COPD. Despite having characteristic symptoms and CT findings as far back as 2017, this patient remained misdiagnosed until incidental trauma imaging prompted reevaluation.

The fact the patient was a lifelong non-smoker with progressive symptoms should have raised suspicion for non–smoking-related structural airway disease. Mislabeling non-smokers with COPD not only delays appropriate care but also exposes patients to unnecessary or ineffective treatments.

This case also illustrates the critical role of radiographic criteria in diagnosis. CT imaging remains the gold standard with diagnostic cutoffs for tracheal and bronchial diameters (>30 mm in men for the trachea; >20 mm right main bronchus; >18 mm left). Associated findings such as bronchiectasis and recurrent infections further support the diagnosis.

Diagnosis is critical because management of MKS is distinct from COPD and hinges on optimizing pulmonary hygiene. Oscillatory positive expiratory pressure devices, airway clearance regimens, vaccination updates, and treatment of recurrent infections are the cornerstones of care. Long-term follow-up is important to monitor for complications, including recurrent pneumonia, progressive bronchiectasis, or respiratory failure.

CONCLUSION

This case illustrates how incidental trauma imaging uncovered a lifelong, but previously unrecognized diagnosis of Mounier-Kuhn syndrome. The patient’s history of chronic cough and recurrent infections in the absence of smoking history underscores the importance of considering structural airway disorders in the differential for “refractory COPD.” Early recognition is crucial as management differs substantially from other chronic airway diseases and focuses on pulmonary hygiene and infection prevention. Greater awareness of MKS can prevent misdiagnosis, optimize patient care, and improve quality of life.