Adult Presentation of Swyer–James–MacLeod Syndrome Masquerading as COPD and Pulmonary Embolism: A Diagnostic Challenge of an Overlooked Childhood Disease - A Case Report

Swyer-James-MacLeod Sendromunun Erişkin Yaşta KOAH ve Pulmoner Emboli ile Karışan Sunumu: Unutulmuş Bir Çocukluk Çağı Hastalığında Tanısal Bir Tuzak - Olgu Sunumu

Sibel Karakaya, Elif Torun Parmaksız, Eylem Tunçay, Metin Karakaya

Abstract

Swyer-James-MacLeod Syndrome (SJMS) is a rare broncho-pulmonary disorder that typically develops following an infection in the lungs during childhood, such as viral bronchiolitis or pneumonia. Radiologically, it is characterized by a unilateral hyperlucent lung, decreased pulmonary vascularity, and air trapping. Clinically, the syndrome can present with symptoms such as cough, sputum production and exertional dyspnea, although it may also remain asymptomatic and be discovered incidentally during imaging. SJMS can mimic various pulmonary conditions, including pulmonary thromboembolism. We present here a case from our outpatient clinic in which a patient was initially evaluated with a preliminary diagnosis of pulmonary thromboembolism, but was ultimately diagnosed with SJMS after thorough clinical and radiological assessment.

Keywords: *Swyer-James MacLeod Syndrome, Unilateral HyperlucentLlung, Air Trapping, Respiratory Bronchiolitis, Bronchiectasis.*

Öz

Swyer-James-MacLeod Sendromu (SJMS), genellikle çocukluk çağında viral bronşiolit veya pnömoni gibi enfeksiyöz bir süreci takiben gelişen nadir bir bronkopulmoner hastalıktır. Radyolojik olarak tek taraflı hiperlusent akciğer, azalmış pulmoner vaskülarite ve hava hapsi ile karakterizedir. Klinik olarak öksürük, balgam ve eforla artan dispne gibi semptomlarla prezente olabilir; ancak asemptomatik de seyrederek görüntüleme sırasında tesadüfen saptanabilir. SJMS, pulmoner tromboemboli (PTE) dahil olmak üzere çeşitli akciğer hastalıklarını taklit edebilir. Bu yazıda, polikliniğimizde PTE ön tanısı ile değerlendirilen, ancak detaylı klinik ve radyolojik değerlendirme sonrası SJMS tanısı konulan bir olgu sunulmaktadır.

Anahtar Kelimeler: *Swyer-James MacLeod Sendromu, Tek Taraflı Hiperlusent Akciğer, Hava Hapsi, Respiratuvar Bronşiolit, Bronşektazi.*

Department of Chest Diseases, University of Health Sciences, Şehit Prof. Dr. İlhan Varank Sancaktepe Training and Research Hospital, İstanbul, Türkiye.

Sağlık Bilimleri Üniversitesi, Şehit Prof. Dr. İlhan Varank Sancaktepe Eğitim ve Araştırma Hastanesi, İstanbul

Submitted (Başvuru tarihi)**:* 26.06.2025 ***Accepted (Kabul tarihi): 13.09.2025

Correspondence (İletişim): Sibel Karakaya, Department of Chest Diseases, University of Health Sciences, Şehit Prof. Dr. İlhan Varank Sancaktepe Training and Research Hospital, İstanbul, Türkiye.

e-mail: celik.sibel.1106@gmail.com

Swyer-James-MacLeod Syndrome (SJMS) is a rare, acquired pulmonary disorder that usually develops after childhood viral bronchiolitis, and most commonly due to adenovirus infection (1,2). It is characterized by obliteration of the small airways, hypoplasia or absence of a pulmonary artery, or the underdevelopment of the peripheral pulmonary vasculature, resulting in a unilateral hyperlucent and hypoperfused lung (1,2). Clinical manifestations range from asymptomatic radiographic findings to chronic respiratory symptoms such as exertional dyspnea, pleuritic chest pain, chronic cough and recurrent lower respiratory tract infections (2,3). Diagnosis is primarily based on characteristic radiological features, especially high-resolution computed tomography (HRCT) and ventilation-perfusion scintigraphy, while pathological confirmation is rarely required (4-7). SJMS is often misdiagnosed as chronic obstructive pulmonary disease (COPD) or pulmonary embolism (PTE) due to their overlapping clinical and radiographic features, leading to inappropriate or delayed treatment (8-10). We present the case of an adult male who undergone treatment for COPD for several years and who was admitted with suspected pulmonary embolism, but was subsequently diagnosed with SJMS following perfusion scintigraphy.

CASE

A 54-year-old male with a 15 pack-year smoking history was referred to our pulmonology outpatient clinic with a 10-day history of right-sided pleuritic chest pain, shoulder discomfort and exertional dyspnea, but no reported hemoptysis or fever. He had been diagnosed with chronic obstructive pulmonary disease (COPD) 5 years earlier based on symptoms of chronic cough and dyspnea, and had been intermittently treated with inhaled bronchodilators. The regimen had included a combination of indacaterol and glycopyrronium bromide, but the patient reported minimal symptomatic improvement, despite long-term use. Of note in his medical history was hospitalization for a lower respiratory tract at the age of 3–4 years, although the exact duration was not documented and no surgical intervention was performed. There was no history of tuberculosis, thromboembolic disease or relevant family history. The patient's vital signs were stable on physical examination, while auscultation revealed end-expiratory wheezing and markedly decreased breath sounds in the left lung. Oxygen saturation was 96% in room air, arterial blood gas: pH: 7.38, PaO_2: 81 mmHg and $PaCO_2$: 37 mmHg.Chest radiography demonstrated right hilar volume loss and decreased vascular markings in the left lung. Laboratory results revealed a D-dimer level of 1489 µg/L, CRP: 1.6 mg/L, and normal troponin and renal function. Pulmonary embolism was initially suspected in the emergency department due to the patient's acute pleuritic chest pain, exertional dyspnea and mildly elevated D-dimer level. Computed tomography pulmonary angiography (CTPA) was thus performed to exclude thromboembolic disease (Figure 1).

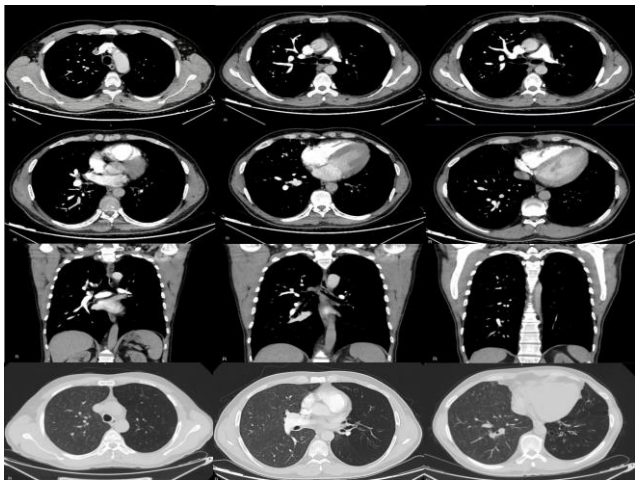

Figure 1: *Computed Tomography Pulmonary Angiography (CTPA) showing all lobes and segments of the right lung with features consistent with respiratory bronchiolitis, while the left lung demonstrates decreased attenuation and reduced broncho-vascular markings. Marked bronchial dilatation, bronchial wall thickening and peribronchial opacities that are particularly prominent in the lower lobe can be observed in both lobes, consistent with bronchiectasis*

The pulmonary arteries, including the main branches, were patent, and no significant thromboembolic event was detected in the distal subsegmental branches. Decreased density with reduced broncho-vascular markings was noted in the left lung, and the scan revealed centrilobular ground-glass opacities in the right lung, consistent with respiratory bronchiolitis, fibroatelectatic bands in the middle lobe and a subpleural air cyst in the right lower lobe. The left lung exhibited reduced volume, diffuses hypodensity, bronchiectasis, bronchial wall thickening and scattered ground-glass opacities that were most prominent in the lower lobe. Pulmonary function tests revealed an obstructive pattern with an FEV_1/FVC ratio of 65%, while transthoracic echocardiog-

raphy *demonstrated* normal biventricular function with no signs of pulmonary hypertension. A perfusion scintigraphy was performed using Tc-99m MAA to further investigate the asymmetrical radiological findings, demonstrating normal perfusion in the right lung, while the left lung showed a global perfusion deficit without segmental or subsegmental defects, consistent with unilateral hypoperfusion (Figure 2).

Based on the combined clinical, radiological and functional findings, the patient was diagnosed with SJMS, a condition likely attributable to his childhood respiratory infection. The initial diagnosis of COPD was reconsidered, and the patient was started on bronchodilator therapy tailored for SJMS-related airflow limitation, with a strong recommendation for smoking cessation and routine vaccinations. The patient reported symptomatic improvement at one-month follow-up. Given the absence of significant functional compromise, he was scheduled for continued outpatient follow-up without need for further intervention.

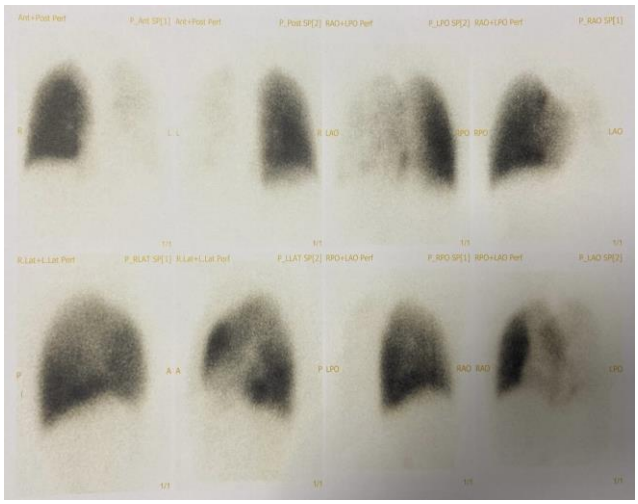

Figure 2: *Perfusion scintigraphy showing normal perfusion in the right lung without significant segmental or subsegmental defects. Perfusion scintigraphy showing absent perfusion in the left lung, and no parenchymal pathology on CT.*

DISCUSSION

Swyer-James-MacLeod Syndrome (SJMS) is a rare pulmonary disorder that was first described in 1953 by Swyer and James. The condition is characterized by unilateral hyperlucent lung, and while it is most frequent in the right lung, it can occasionally present with bilateral involvement (11). The pathogenesis involves post-infectious bronchiolitis obliterans, most commonly due to adenovirus subtypes B21, B3 or B7 in childhood, resulting in small airway obstruction and vascular hypoplasia (1,2). The history of severe lower respiratory tract infection during early life noted in our patient is a well-established risk factor for SJMS (2,3). In the original cases described by Swyer and James, spirometry typically demonstrated airflow obstruction associated with features of bronchiolitis obliterans (11), although there is evidence that SJMS may also be linked to the development of pulmonary hypertension in later stages of life (6,7). It should be kept in mind that SJMS can mimic acute pulmonary embolism (PTE), both clinically and radiographically. The sudden-onset dyspnea, pleuritic chest pain and hypoxemia symptoms common to PTE overlap with SJMS in patients with significant unilateral perfusion abnormalities. However, while PTE is an acute vascular event requiring immediate intervention, SJMS is a chronic condition with congenital or acquired pulmonary vascular underdevelopment. Differentiating between the two is essential, as misdiagnosis may lead to unnecessary anticoagulation or delayed appropriate management (9,10). In our case, the patient's acute chest symptoms and elevated D-dimer level initially raised suspicion for PTE, but further evaluation, including perfusion scintigraphy, pointed to the correct diagnosis. Another diagnostic pitfall is the misclassification of SJMS as COPD. Due to the presence of chronic cough, dyspnea and airflow limitation, patients with SJMS are often empirically treated for COPD without adequate imaging or pulmonary function evaluation. In our patient, bronchodilator therapy had been prescribed for presumed COPD for several years, but the lack of significant clinical improvement prompted further investigation. The spirometric pattern in SJMS often shows an obstructive component that, unlike COPD, is typically unilateral and associated with matched radiographic findings of lung volume loss, vascular attenuation and air trapping. Moreover, the progressive nature and systemic manifestations seen in classic COPD are usually absent in cases of SJMS. Previously reported cases have described patients treated for COPD for years until HRCT revealed unilateral hyperlucency and vascular hypoplasia consistent with SJMS (8), underscoring the importance of detailed history-taking and appropriate imaging in patients with atypical or treatment-refractory obstructive symptoms. HRCT remains the cornerstone of structural assessment, typically showing hyperlucency,

air trapping, bronchiectasis and vascular paucity in the affected lung. Ventilation-perfusion scintigraphy provides complementary functional information, characteristically demonstrating unilateral global hypoperfusion without segmental defects, a finding that helps distinguish SJMS from pulmonary embolism (6,7,9). The combined use of HRCT and scintigraphy can thus enhance diagnostic accuracy and prevent misinterpretation. The management of SJMS is usually conservative, tailored to symptom control and the prevention of complications. Treatments include inhaled bronchodilators, vaccination against influenza and pneumococcus, smoking cessation and pulmonary rehabilitation. Patients should also be followed for potential long-term complications such as recurrent pulmonary infections, hemoptysis and the gradual development of pulmonary hypertension (6,7). In selected cases with localized disease and recurrent infections, surgical interventions such as lobectomy may be considered (7). Our patient responded well to conservative measures with symptomatic improvement at follow-up.

CONCLUSION

Swyer-James-MacLeod Syndrome (SJMS) is a rare but important differential diagnosis in adults presenting with unilateral hyperlucent lung, chronic respiratory symptoms or perfusion abnormalities suggestive of pulmonary embolism. The presented case emphasizes the importance of a detailed history, advanced imaging (HRCT and perfusion scintigraphy) and pulmonary function tests for the establishment of a correct diagnosis. Clinicians should keep SJMS in mind when encountering patients with a history of lower respiratory infections in childhood and inadequate response to standard COPD therapy. Our report highlights the role of perfusion scintigraphy and HRCT in the diagnosis of this often-overlooked condition and in the prevention of misdiagnoses.

CONFLICTS OF INTEREST

None declared.

AUTHOR CONTRIBUTIONS

Concept - S.K., E.T.P., E.T, M.K.; Planning and Design - S.K., E.T.P., E.T, M.K.; Supervision - E.T.P., E.T.; Funding - S.K., E.T.P., E.T, M.K.; Materials - S.K., E.T.P.; Data Collection and/or Processing - E.T.P., E.T, M.K.; Analysis and/or Interpretation - S.K., E.T.P.; Literature Review - S.K., E.T.P., M.K.; Writing - S.K., E.T.P.; Critical Review - E.T.P., E.T, M.K.

REFERENCES

1. Shapiro SD, Snider GL, Rennard SI. Chronic Bronchitis and Emphysema. In: Murray JF, Nadel JA, eds. Textbook of Respiratory Medicine. Philadelphia: Elsevier Saunders; 2005:1115-67.
2. Capela C, Gouveia P, Sousa M, Regadas MJ. Adult Diagnosis of Swyer-James-Macleod Syndrome: A Case Report. J Med Case Rep 2011; 5:2. [CrossRef]
3. Bernardi F, Cazzato S, Poletti V, Tassinari D, Burnaccini M, Zompatori M, et al. Swyer-James Syndrome: Bronchoalveolar Lavage Findings in two Patients. Eur Respir J 1995; 8:654-7. [CrossRef]
4. Conti L, Palmieri G, Delfanti R, Grassi C, Daccò MD, Capelli P. Swyer-James-MacLeod Syndrome Presenting as Spontaneous Pneumothorax in An Adult: Case Report and Literature Review. Radiol Case Rep 2021; 16:1133-7. [CrossRef]
5. Hansell DM, Bankier AA, MacMahon H, McLoud TC, Müller NL, Remy J. Fleischner Society: Glossary of Terms for Thoracic Imaging. Radiology 2008; 246:697-722. [CrossRef]
6. Behrendt A, Lee Y. Swyer-James-MacLeod Syndrome. In: StatPearls. Treasure Island (FL): StatPearls Publishing; 2025 Jan.
7. Lee EY, Boiselle PM, Cleveland RH. Multidetector CT Evaluation of Congenital Lung Anomalies. Radiology 2008; 247:632-48. [CrossRef]
8. Intepe YS, Yuksel M, Tanyeri B. Swyer-James-MacLeod Syndrome Misdiagnosed as COPD: A Case Report. Kuwait Med J 2017; 49:335-9.
9. Arslan N, Ilgan S, Ozkan M, Yuksekol I, Bulakbasi N, Pabuscu Y, et al. Utility of Ventilation and Perfu-sion Scan in the Diagnosis of Young Military Rec-ruits with an Incidental Finding of Hyperlucent Lung. Nucl Med Commun 2001; 22:525-30. [CrossRef]
10. Lema GD, Melaku EE, Tekle HA, Aydagnuhm GB, Gebeyaw ED. Swyer-James-Macleod Syndrome Mimicking Pulmonary Embolism in Adults: A Case Report. J Med Case Rep 2024; 18:359.[CrossRef]
11. Swyer PR, James GCW. A Case of Unilateral Pulmonary Emphysema. Thorax 1953; 8:133-6.