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A Case of Sporadic Pulmonary Lymphangioleiomyomatosis in a Woman of Reproductive Age Exacerbated by Pregnancy

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Abstract

Lymphangioleiomyomatosis (LAM) is a rare and progressive multisystem neoplastic disorder that results from the pathological infiltration of abnormal smooth muscle–like LAM cells into the pulmonary interstitium, lymphatic vessels, and abdominopelvic organs. These LAM cells demonstrate inappropriate proliferation and migration as well as heightened proteolytic activity due to dysregulation of the mammalian target of rapamycin (mTOR) signalling pathway [1]. Clinically, patients often present with exertional dyspnoea, recurrent pneumothorax, progressive airflow limitation, or extrapulmonary features such as chylous effusions, lymphadenopathy, or renal angiomyolipomas [2]. In this case report, we describe a 30-year-old woman who developed progressive dyspnoea during pregnancy, with persistence of symptoms into the postpartum period, and subsequently developed painless cervical lymphadenopathy. High-resolution computed tomography (HRCT) of the chest revealed widespread thin-walled pulmonary cysts consistent with LAM, while pulmonary function testing showed preserved spirometry with pronounced gas trapping and a moderately reduced diffusing capacity. Laboratory investigations demonstrated no evidence of autoimmune disease, infection, or malignancy. There were no clinical or radiological features suggestive of tuberous sclerosis complex (TSC). A diagnosis of sporadic pulmonary LAM was therefore established. The patient was managed conservatively with smoking cessation, avoidance of exogenous oestrogen, pneumothorax counselling, and structured long-term radiological and physiological surveillance. This case highlights the importance of recognizing LAM in young women and illustrates how pregnancy may unmask or accelerate disease through hormonal mechanisms [3].

Keywords: Lymphangioleiomyomatosis (LAM); TSC1 or TSC2; Migration; Sporadic pulmonary LAM

Introduction

Lymphangioleiomyomatosis is a rare cystic lung disease characterized by the proliferation of abnormal smooth muscle–like LAM cells that progressively infiltrate and remodel lung parenchyma, lymphatic vessels, and abdominal organs [1]. This pathological process occurs due to mutations in the tumour suppressor genes TSC1 or TSC2, which normally encode regulatory proteins (hamartin and tuberin) that modulate mTORC1 activity. Mutations in these genes result in constitutive activation of the mTOR pathway, leading to uncontrolled LAM cell proliferation, migration, and secretion of tissue-degrading proteases [4]. LAM occurs in two clinically recognised forms. Sporadic LAM (S-LAM) occurs in women without clinical features of TSC, whereas TSC-associated LAM (TSC-LAM) occurs in patients with tuberous sclerosis complex and is often diagnosed earlier in life with more extensive extrapulmonary involvement [4].

LAM overwhelmingly affects women, particularly during their reproductive years, highlighting the influence of hormonal factors, especially oestrogen on disease activity [3]. Pregnancy and exogenous oestrogen exposure have both been reported to accelerate disease progression through enhanced survival and proliferation of LAM cells [3,5]. Common manifestations include exertional dyspnoea, recurrent spontaneous pneumothorax, airflow obstruction, persistent cough, chylous effusions, and renal

angiomyolipomas [2,6]. Diagnosis is heavily reliant on HRCT, which typically demonstrates diffuse, thin-walled, round or ovoid pulmonary cysts evenly distributed throughout relatively normal parenchyma.5 Serum VEGF-D levels above 800 pg/mL strongly support the diagnosis and may obviate the need for lung biopsy, although this biomarker is not universally available [7]. This case describes sporadic pulmonary LAM in a young woman whose pregnancy likely precipitated or accelerated symptom onset, providing insight into hormonal influences, diagnostic considerations, and evidence-based management strategies.

Case Report

A 30-year-old woman presented with progressively worsening shortness of breath which began during the third trimester of her first pregnancy and persisted for eight months postpartum. She described a long-standing intermittent dry cough over several years, which became more noticeable during late pregnancy. She also reported transient dizziness and the development of painless cervical lymphadenopathy. She denied haemoptysis, pleuritic pain, fevers, weight loss, or symptoms suggestive of pneumothorax.

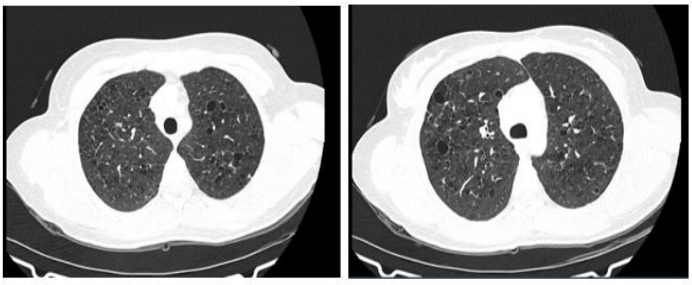

Figure 1: *HRCT showing bilateral cysts of varying sizes in upper zone of lungs, some in close proximity to pleura.*

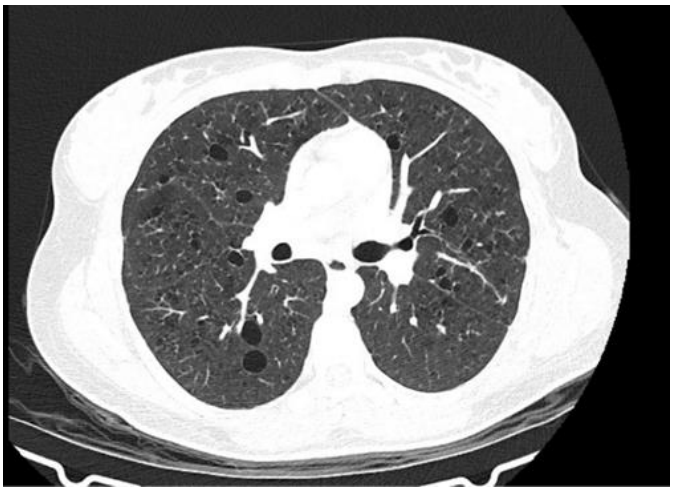

Figure 2: *HRCT showing cysts of varying sizes in middle zone of bilateral lung fields.*

Her medical history included childhood pneumonia but no chronic pulmonary disease. There were no features suggestive of tuberous sclerosis, including seizures, dermatological lesions, or renal abnormalities. She had a 15–pack-year history of cigarette smoking and daily marijuana/THC use, and she worked as a cleaner with exposure to aerosols and volatile chemicals. She denied exposures to birds, moulds, or other antigens known to cause hypersensitivity pneumonitis. She also denied symptoms of connective tissue disease. Her family history was notable only for a relative with lung cancer. On examination, the patient was comfortable, with normal respiratory effort and an oxygen saturation of 98% on room air. She exhibited no clubbing, cyanosis, lymphoedema, or cutaneous manifestations of TSC. Respiratory auscultation revealed clear vesicular breath sounds bilaterally without wheeze or crackles. Cardiovascular, abdominal, and neurological examinations were normal.

Investigations

Radiological findings

A CT scan of the chest, abdomen, and pelvis that was performed to investigate lymphadenopathy demonstrated multiple thin-walled pulmonary cysts throughout both lungs, ranging from a few millimetres to 2 cm in diameter. These cysts were diffuse and evenly distributed, particularly within the upper and mid lung zones. Mild hilar and axillary lymphadenopathy was noted. There was no consolidation, pleural effusion, bronchiectasis, or pneumothorax. A dedicated HRCT confirmed widespread thin-walled cysts characteristic of LAM.5 There were no nodules, irregularly shaped cysts, or interstitial infiltrates to suggest pulmonary Langerhans cell histiocytosis. No radiological features suggested Birt–Hogg–Dubé syndrome, lymphoid interstitial pneumonia, or emphysema (Figures 1-3).

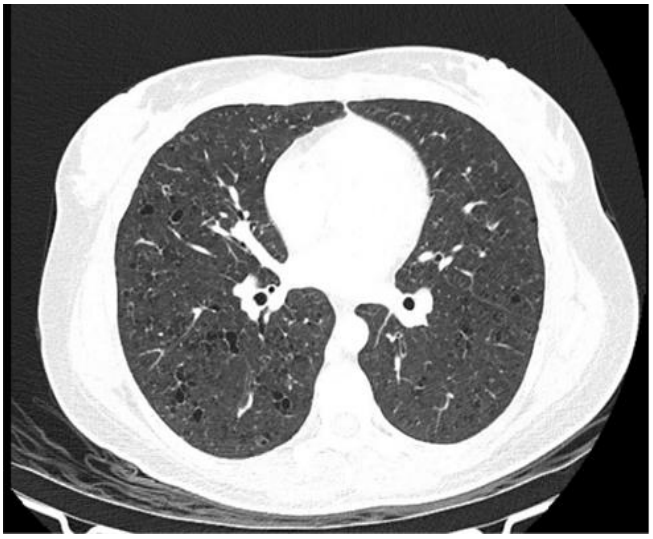

Figure 3: *HRCT showing small cysts in lower zones of the lungs.*

Pulmonary function testing

Spirometry showed preserved volumes with an FEV_1 of 87% predicted and an FVC of 91.8% predicted. The FEV_1/FVC ratio was preserved. Lung volumes revealed a normal total lung capacity (100.6% predicted) and a markedly elevated RV/TLC ratio (156%), indicating substantial gas trapping. The DLCO was moderately reduced at 63.7% predicted and the KCO was 72.2% predicted. This physiological pattern is typical of early LAM, in which cystic destruction precedes significant airflow limitation [1].

Laboratory tests

Laboratory investigations revealed a weakly positive ANA of 1:40 but no ENA antibodies or rheumatoid factor. ESR was mildly elevated, but CRP was normal. Tumour markers were within normal limits. Blood counts, renal and liver function tests, serum immunoglobulins, electrolytes, and infectious disease screening (HIV, hepatitis B and C) were unremarkable. VEGF-D testing was unavailable locally and declined privately due to cost [7].

Abdominal imaging

A complete abdominal ultrasound revealed normal kidneys with no angiomyolipomas or cysts. Other abdominal organs were normal.

Diagnostic Conclusion

The diagnosis of sporadic pulmonary LAM was made based on the classic HRCT findings, physiological evidence of gas trapping and impaired diffusion, and the absence of clinical or radiological signs of TSC. Pregnancy likely triggered or accelerated disease onset due to hormonal influences.3 Differentials including pulmonary Langerhans cell histiocytosis, emphysema, lymphoid interstitial pneumonia, Sjögren-associated cystic disease, and Birt–Hogg–Dubé syndrome were excluded clinically and radiologically [5].

Management

The patient received counselling regarding smoking and marijuana cessation, avoidance of oestrogen-containing contraception, and early recognition of pneumothorax symptoms. In accordance with current guidelines, sirolimus was not commenced because her lung function was stable and there was no radiological evidence of progression [8]. A structured surveillance plan was implemented consisting of HRCT imaging every one to two years, annual pulmonary function tests, and periodic renal imaging every two to three years to screen for late-developing angiomyolipomas [4,7]. At the six-month review, symptoms had improved postpartum and lung function remained stable.

Discussion

LAM is a multisystem disease driven by mTOR-mediated LAM cell proliferation, lymphangiogenesis, and protease-driven cystic destruction [1]. Its strong hormonal sensitivity explains why pregnancy may worsen symptoms and accelerate disease progression [3,6]. HRCT is central to diagnosis due to its ability to detect characteristic cystic parenchymal destruction. Serum VEGF-D, when available, provides a non-invasive diagnostic biomarker that may avoid the need for lung biopsy [7]. mTOR inhibition with sirolimus has revolutionised management, stabilising lung function, reducing chylous effusions, and shrinking lymphatic masses [8]. Conservative management is appropriate in clinically stable individuals, particularly those whose symptoms emerge during pregnancy rather than showing progressive decline. Ongoing follow-up remains essential as disease progression may vary widely among patients.

Conclusion

This case underscores the importance of considering sporadic pulmonary LAM in young women presenting with dyspnoea, particularly when symptoms arise or worsen during pregnancy. HRCT was crucial in establishing the diagnosis in the absence of VEGF-D testing. Comprehensive evaluation excluded TSC and other cystic lung diseases. Conservative management, avoidance of hormonal triggers, and structured radiological and functional surveillance were appropriate given the patient's stable clinical course. Early recognition and long-term monitoring are essential to optimise outcomes in LAM.

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