CASE REPORT

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Pulmonary Alveolar Microlithiasis: Bedside Lung and Diaphragmatic Ultrasound Findings in Stage III Disease

Pulmoner Alveoler Mikrolitiyazis: Evre III Hastalıkta Yatak Başı Akciğer ve Diyafragma Ultrasonografi Bulguları

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ABSTRACT

Pulmonary alveolar microlithiasis (PAM) is an extremely rare autosomal recessive disorder caused by *SLC34A2* mutations, leading to intra-alveolar calcium-phosphate microlith accumulation. To date, lung ultrasound (LUS) findings in PAM have been reported in only a single case report. We present a 35-year-old male with genetically confirmed stage III PAM who presented with acute hypoxemic respiratory failure. Bedside LUS revealed marked pleural irregularity and thickening (total 5.6 mm; parietal 4.4 mm, visceral 1.2 mm) and complete absence of B-lines despite extensive ground-glass opacities on computed tomography. Diaphragmatic ultrasound showed a thickening fraction of 31.5%, indicating preserved contractile reserve. Arterial blood gas on room air (PaO_2 48 mmHg), normal infection markers, negative D-dimer (0.3 µg/mL), and echocardiography showing no acute right heart strain effectively excluded alternative causes of acute hypoxemia. The absence of B-lines may be attributed to an acoustic barrier created by subpleural microliths and microcysts. Preserved diaphragmatic function suggests that hypoxemia arose from gas-exchange impairment rather than ventilatory pump failure. Given its extreme rarity, this second ever description of LUS in PAM confirms that bedside ultrasound may serve as a radiation-free monitoring tool in this orphan disease.

Keywords: Pulmonary alveolar microlithiasis, lung ultrasound, diaphragm, B-lines, pleural thickening, respiratory failure, *SLC34A2* gene

ÖZ

Pulmoner alveoler mikrolitiyazis (PAM), *SLC34A2* gen mutasyonlarına bağlı son derece nadir görülen otozomal resesif bir hastalık olup alveol boşluklarında kalsiyum-fosfat mikrolitlerinin birikimi ile karakterizedir. Bugüne kadar PAM'da akciğer ultrasonografisi (USG) bulguları literatürde yalnızca tek bir olguda bildirilmiştir. Bu yazıda, genetik olarak doğrulanmış evre III PAM tanılı, akut hipoksemik solunum yetmezliği ile başvuran 35 yaşında bir erkek hasta sunulmaktadır. Yatak başı akciğer USG'de belirgin plevral düzensizlik ve kalınlaşma (toplam 5,6 mm; paryetal 4,4 mm, viseral 1,2 mm) ve bilgisayarlı tomografi'de yaygın buzlu cam dansitelerine rağmen B-çizgileri saptanmadı. Diyafragma USG'de kalınlaşma fraksiyonu %31,5 olarak ölçüldü; bu değer korunmuş kontraktil rezervi göstermektedir. Oda havasında arter kan gazı (PaO_2 48 mmHg), normal enfeksiyon belirteçleri, negatif D-dimer (0,3 µg/mL) ve ekokardiyografide akut sağ kalp yüklenme bulgusu olmaması enfeksiyon, pulmoner emboli ve kardiyak disfonksiyon gibi akut hipoksemi nedenlerini dışlamıştır. B-çizgilerinin olmaması, subplevral mikrolitlerin ve mikrokistlerin oluşturduğu akustik bariyerden kaynaklandığı düşünülmektedir. Korunmuş diyafragma fonksiyonu, hipokseminin gaz değişim bozukluğundan kaynaklandığını düşündürmektedir. Yatak başı USG nadir görülen bu hastalıkta radyasyon içermeyen bir izlem aracı olarak kullanılabilir.

Anahtar Kelimeler: Pulmoner alveoler mikrolitiyazis, akciğer ultrasonografisi, diyafragma, B-çizgileri, plevral kalınlaşma, solunum yetmezliği, *SLC34A2* geni

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INTRODUCTION

Pulmonary alveolar microlithiasis (PAM) is an exceedingly rare, autosomal recessive genetic lung disease characterized by the diffuse accumulation of calcium-phosphate microliths within the alveolar spaces[1]. This condition is primarily caused by inactivating mutations in the *SLC34A2* gene, which encodes the type IIb sodium-phosphate cotransporter in alveolar type II cells[2]. The resulting defect in phosphate clearance leads to intra-alveolar stone formation, progressively impairing gas exchange and leading to pulmonary fibrosis, respiratory failure, and cor pulmonale[1,2].

A hallmark of PAM is the clinico-radiological dissociation, where extensive "sandstorm-like" micronodular patterns on imaging are found in patients with relatively mild or no symptoms for decades[1]. While computed tomography (CT) remains the most useful test for diagnosis and staging, the role of lung ultrasound (LUS) in PAM is documented by only a single case report, which described the sonographic pleural abnormalities in PAM[3,4]. This report aims to expand the sonographic literature by presenting LUS and diaphragmatic ultrasound (DUS) findings in a patient with stage III PAM.

CASE REPORT

A 35-year-old male with a 16-year history of PAM presented to the emergency department with a two-day history of progressively worsening dyspnea and acute-onset hypoxemia. The diagnosis of PAM had been established 16 years earlier based on typical symptoms and high-resolution CT findings. Genetic testing performed at that time confirmed a homozygous inactivating mutation in the *SLC34A2* gene. Over the preceding decade, he had gradual exertional breathlessness, but his dyspnea acutely worsened in the last 48 hours. There was no associated fever, productive cough, chest pain, or hemoptysis. He reported compliance with domiciliary oxygen therapy only intermittently and had never smoked. His family history was unremarkable for pulmonary disease.

On arrival, the patient was tachypneic. Vital signs were as follows: blood pressure 118/76 mmHg, heart rate 102 bpm, temperature 36.7 °C, respiratory rate 26 breaths/min, and peripheral oxygen saturation (SpO_2) 79% on room air. Chest auscultation revealed diffusely audible rhonchi without crackles or wheezing. There was no peripheral edema, jugular venous distension, or clinical evidence of right-sided heart failure.

Arterial blood gas (ABG) analysis on room air revealed: pH 7.38, $PaCO_2$ 42 mmHg, PaO_2 48 mmHg, and HCO_3^- 24 mEq/L. Complete blood count showed leukocytosis (12.4×10^9/L); C-reactive protein was 8 mg/L, and procalcitonin was 0.1 ng/mL, making infection unlikely. Cardiac evaluation included high-sensitivity troponin T (<3 ng/L) and a 12-lead electrocardiography showing sinus tachycardia without ischemic changes or right heart strain. A D-dimer level was 0.3 μg/mL, effectively excluding pulmonary embolism as a cause of acute hypoxemia. A Wells score for pulmonary embolism was 1.5 (low probability).

Immediately after the initiation of supplemental oxygen, a bedside 12-point LUS was performed by an emergency medicine specialist with 7 years of experience in LUS. All ultrasound examinations were performed using SonoHealth® D2CP with high-frequency linear (5-12 MHz) and phased-array probes (SonoHealth Medical Technologies, Guangzhou, China). The 12-point LUS protocol involved systematic scanning of the anterior, lateral, and posterior chest walls, dividing each hemithorax into six zones: anterior-superior, anterior-inferior, lateral-superior, lateral-inferior, posterior-superior, and posterior-inferior. Each zone was scanned in both longitudinal and transverse planes using a standardized approach, with the transducer placed perpendicular to the ribs to avoid acoustic shadowing. Three consecutive respiratory cycles were recorded for each measurement, and the average value was reported. Bilateral anterior-superior, posterior-superior, and posterior-inferior zones were systematically examined with the patient in a semi-recumbent position. The pleural line was markedly thickened and irregular in all evaluated regions. Using the linear probe, total pleural thickness measured 5.6 mm (average of three measurements: 5.5, 5.7, 5.6 mm), comprising a parietal layer of 4.4 mm (4.3, 4.5, 4.4 mm) (Figure 1a) and a visceral layer of 1.2 mm (1.1, 1.3, 1.2 mm) (Figure 1b). The visceral pleura appeared hyperechoic, fragmented, and irregular (Figure 1b and c). No vertical B-lines or comet-tail reverberation artifacts were observed.

DUS was performed using the phased-array probe positioned in the right mid-axillary line, between the 8th and 10th intercostal spaces. The diaphragm was visualized in the zone of apposition, and M-mode was used to capture respiratory-phase thicknesses. End-expiratory thickness was 1.9 mm, and end-inspiratory thickness increased to 2.5 mm. The diaphragmatic thickening fraction was calculated as $[(2.5\text{-}1.9)/1.9] \times 100 = 31.5\%$. Diaphragmatic excursion was visually preserved.

A postero-anterior chest radiograph demonstrated the classic "sandstorm" appearance: diffuse, bilateral, fine micronodular opacities with a ground-glass density, more pronounced in the lower zones (Figure 2a).

As the bedside LUS and portable chest radiograph revealed no evidence of consolidation, cardiogenic pulmonary edema, pleural effusion, or pneumothorax that could otherwise account for the acute deterioration, a thoracic CT scan was obtained to exclude occult parenchymal complications and better delineate the underlying disease status. The scan confirmed symmetrical, dense alveolar microliths distributed predominantly in the posterior and inferior lung segments,

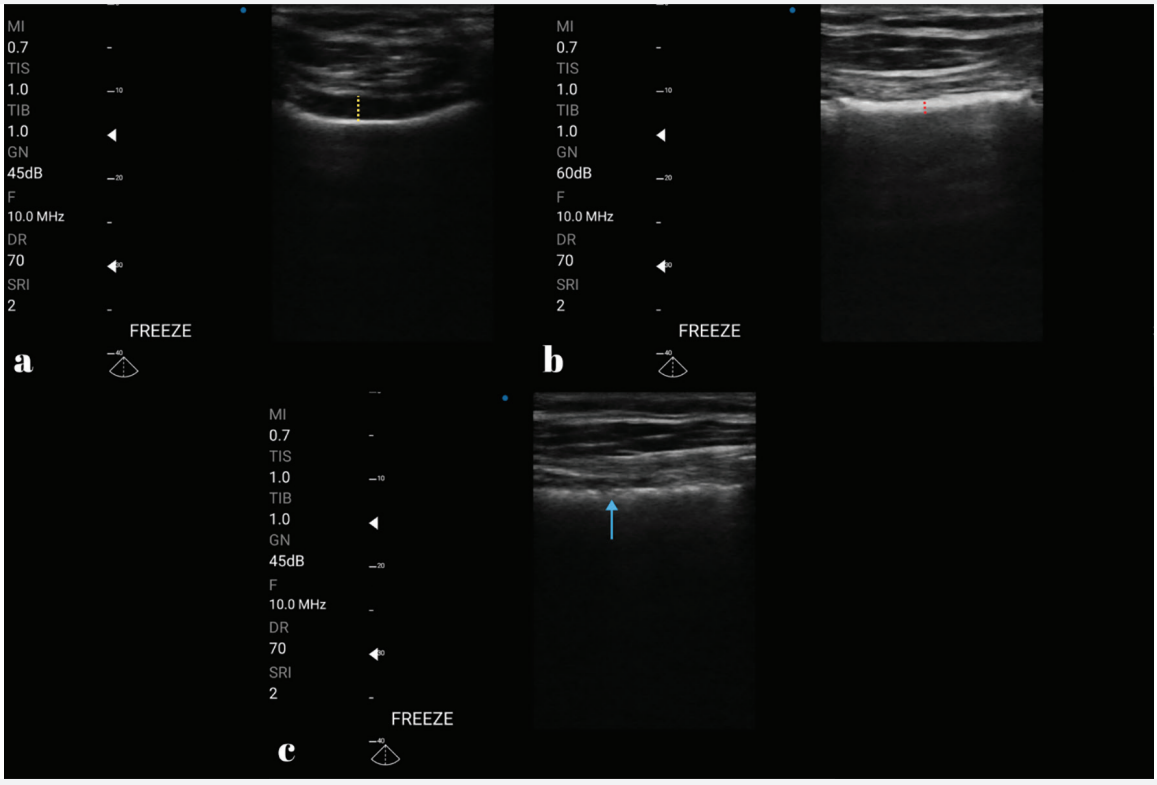

Figure 1. High frequency linear probe LUS of the right anterior superior zone (mid clavicular line, second intercostal space, longitudinal orientation). (a) Markedly thickened parietal pleura (4.4 mm, dashed yellow line). (b) Hyperechoic, thickened visceral visceral pleura (1.2 mm, dashed red line). (c) Hyperechoic fragmented visceral pleural layer

LUS: Lung ultrasound

accompanied by interlobular septal thickening, consistent with a "crazy-paving" pattern, and multiple subpleural traction cysts (Figure 2b). Per established staging criteria[2], the radiographic "sandstorm" appearance with partial obliteration of the lower mediastinal and diaphragmatic borders, alongside these High-resolution CT findings of progressive interstitial fibrosis, are consistent with advanced stage III PAM.

The patient was treated with oxygen 4-6 L/min via mask, inhaled salbutamol and ipratropium, and intravenous hydration. His dyspnea improved, and SpO_2 stabilized at 91-93% on 3 L/min of oxygen. Repeat ABG after 6 hours showed pH 7.41, $PaCO_2$ 40 mmHg, PaO_2 68 mmHg, HCO_3^- 25 mEq/L. He was admitted to the pulmonology ward for further observation and multidisciplinary evaluation, including lung-transplant candidacy assessment, and was discharged home on domiciliary oxygen after five days of follow-up.

Written informed consent was obtained from the patient for publication of this case report and accompanying images. The study was conducted in accordance with the Declaration of Helsinki.

DISCUSSION

This case details the bedside LUS and DUS findings in a patient with stage III PAM. The sonographic appearance of PAM has rarely been reported, with only one prior description[4]. Our observations corroborate their findings of a thickened, irregular hyperechoic pleural line, while offering assessment of diaphragmatic function in PAM.

Rea et al.[4] reported pleural line thicknesses between 1.7 mm and 2.9 mm depending on the transducer used. In our patient, total pleural thickness measured 5.6 mm, comprising 4.4 mm of parietal and 1.2 mm of visceral pleura, both well above the 3 mm threshold considered pathological[5]. This degree of thickening supports the concept that the calcific and inflammatory process of PAM can involve pleura at stage III, paralleling the subpleural interstitial changes and microcysts frequently seen on CT[3,4].

An important observation in both cases is the absence of B-lines despite extensive parenchymal calcifications and ground-glass opacities on CT. In most interstitial lung diseases, such opacities generate vertical comet-tail artifacts due to acoustic

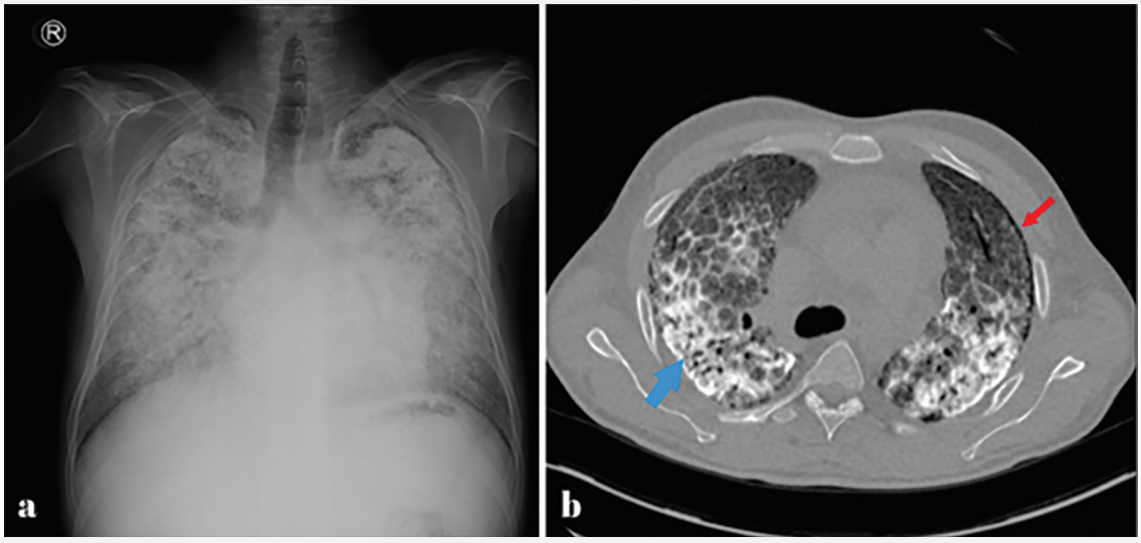

Figure 2. Radiological findings in pulmonary alveolar microlithiasis. (a) Posteroanterior chest radiograph showing diffuse sand like micronodular calcifications. (b) Axial thoracic CT image revealing dense alveolar microliths (blue arrow), air space obliteration, and subpleural cystic changes (red arrow), predominantly in the inferoposterior regions

CT: Computed tomography

impedance mismatches at the air–tissue interface[6]. Rea et al.[4] proposed that subpleural microliths and microcystic changes create a dense acoustic barrier that reflects ultrasound waves, preventing deeper penetration and artifact formation. Our case supports this finding.

In PAM, chronic hypoxia and restrictive physiology increase the work of breathing and may eventually lead to cor pulmonale[1]. Rea et al.[4] observed reduced pleural line motion during respiration, attributing it to parenchymal stiffness and obesity, but did not quantify diaphragmatic function. We measured the TFdi at approximately 31.5%. Although this value lies slightly below the >36% threshold proposed for healthy individuals and the mean of 36.8±6.5% reported in successfully weaned patients, it remains above the cut-offs associated with diaphragmatic dysfunction: <30% for weaning failure and <20% for severe weakness[7-10]. Values in the 15-30% range are considered indicative of adequate activation, placing our patient's measurement within the preserved contractile reserve spectrum[9]. The measurement was obtained during spontaneous breathing without positive pressure ventilation, conditions that yield the most reliable indices[7]. Our patient presented with a room-air SpO_2 of 79%, may suggest that the respiratory distress originated primarily from gas-exchange impairment due to alveolar filling rather than from ventilatory pump failure. The novelty of combining LUS and DUS in PAM lies in its potential to simultaneously assess disease severity at the pleural level and respiratory muscle function, without the need for ionizing radiation. This dual approach may be particularly valuable for monitoring disease progression and assessing functional reserve in PAM patients being evaluated for lung transplantation.

Currently, no proven disease-modifying therapy exists for PAM. Supportive measures include long-term oxygen therapy to maintain SpO_2 ≥88% (or >90% if cor pulmonale develops), and improved gas exchange with nasal continuous positive airway pressure has been described in a single case[1,3]. Disodium etidronate has been used to inhibit hydroxyapatite crystal formation; although radiological and symptomatic improvements have been reported, efficacy in adults remains uncertain[1,2]. Dietary phosphate restriction lacks robust human data, though often advised[3]. Vaccination against influenza, pneumococcus, and coronavirus disease 2019, along with smoking cessation, is recommended[3]. Lung transplantation remains the only definitive treatment, and disease recurrence in the graft has not yet been reported[2,3]. Transplant timing is guided by right-heart failure or severe respiratory failure[2].

Several limitations must be acknowledged. This is a single case report, requiring validation in larger cohorts. The influence of body habitus and inter-operator variability on measurements could not be systematically assessed as all measurements were performed by a single operator. Additionally, the TFdi cut-offs cited derive mainly from critical care populations; disease-specific reference ranges for chronic restrictive disorders are not established.

CONCLUSION

In conclusion, this case suggests that stage III PAM may present with pleural thickening and the absence of B-lines. In this patient, preserved diaphragmatic function suggests that hypoxemia may be due to gas exchange failure, without evidence of respiratory muscle insufficiency. Bedside ultrasound may be useful as a radiation-free tool for monitoring disease progression in PAM.

Ethics

Informed Consent: Written informed consent was obtained from the patient for publication of this case report and accompanying images. The study was conducted in accordance with the Declaration of Helsinki.

Footnotes

Authorship Contributions

Surgical and Medical Practices: K.Y., A.S.G., Concept: K.Y., M.K.A., Ö.J., A.S.G., Design: K.Y., M.K.A., Ö.J., A.S.G., Data Collection or Processing: K.Y., M.K.A., Ö.J., A.S.G., Analysis or Interpretation: K.Y., M.K.A., Ö.J., A.S.G., Literature Search: Ö.J., A.S.G., Writing: K.Y., M.K.A.

Conflict of Interest: No conflict of interest was declared by the authors.

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