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Pulmonary Alveolar Microlithiasis Diagnosed by High-Resolution Computed Tomography, Bronchoalveolar Lavage, and Genetic Testing in a Young Adult

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Abstract

We report the case of a 19-year-old Saudi man who presented with a four-day history of cough, whitish sputum, and shortness of breath. Despite relatively mild symptoms and preserved oxygen saturation, chest imaging showed extensive bilateral pulmonary calcification. High-resolution computed tomography demonstrated diffuse sand-like calcified micronodules, interlobular septal thickening with a crazy-paving pattern, and the black pleura sign. Bronchoalveolar lavage cytology revealed microliths with multinucleated giant cells, while genetic testing identified a pathogenic variant in the *SLC34A2* gene, supporting the diagnosis. In this patient, the relatively mild clinical presentation contrasted with the extensive radiological abnormalities. The complementary findings from imaging, bronchoalveolar lavage cytology, and the available genetic result supported the diagnosis without the need for surgical lung biopsy. Following completion of the diagnostic workup, the patient was referred for lung transplantation evaluation, and genetic screening of family members was recommended.

Categories: Pulmonology

Keywords: bronchoalveolar lavage, diffuse pulmonary calcification, high-resolution computed tomography, pulmonary alveolar microlithiasis, slc34a2

Introduction

Pulmonary alveolar microlithiasis (PAM) is a rare inherited lung disease, usually transmitted in an autosomal recessive pattern, characterized by the gradual accumulation of calcium phosphate microliths within the alveolar spaces [1]. It is caused by pathogenic variants in the *SLC34A2* gene, which encodes a sodium phosphate cotransporter expressed mainly in type II alveolar epithelial cells. In this autosomal recessive disorder, disease-causing variants generally affect both alleles of *SLC34A2*, resulting in impaired phosphate transport and progressive deposition of calcium phosphate material within the lungs [2].

A characteristic feature of PAM is the striking discrepancy between the extent of radiological abnormalities and the relatively mild clinical symptoms, particularly in the early stages of the disease [3]. With progression, patients may develop pulmonary fibrosis, pulmonary hypertension, cor pulmonale, and respiratory failure [4]. High-resolution computed tomography (HRCT) typically demonstrates diffuse calcified micronodules, often associated with interlobular septal thickening, a crazy-paving appearance, and the characteristic black pleura sign [5].

Diagnosis is usually suggested by characteristic imaging findings and may be supported by the identification of microliths in bronchoalveolar lavage fluid. Genetic testing can provide diagnostic confirmation through identification of pathogenic *SLC34A2* variants [2,5,6]. This report describes a young adult with PAM who had experienced a previous undiagnosed respiratory episode and was subsequently diagnosed through the combined use of HRCT, bronchoalveolar lavage cytology, and genetic testing. The case highlights the importance of recognizing the characteristic clinico-radiological pattern and demonstrates how a multimodal diagnostic approach can establish the diagnosis without the need for surgical lung biopsy.

Case Presentation

A 19-year-old Saudi man was admitted in October 2023 with a four-day history of cough, a small amount of whitish sputum, and shortness of breath. He reported a similar respiratory episode four years earlier that had remained undiagnosed. He was a non-smoker with no history of occupational dust exposure, bird exposure, or known allergies. Family history relevant to PAM and parental consanguinity were not documented and were therefore considered unknown.

On examination, he had an average body build and a pigeon-shaped chest. His vital signs were stable, with an oxygen saturation of 96% on room air, heart rate of 74 beats/minute, respiratory rate of 15

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breaths/minute, and blood pressure of 118/70 mmHg. Cardiac examination was unremarkable. Chest auscultation revealed scattered bilateral wheezes. There was no cyanosis, clubbing, or peripheral edema.

Laboratory investigations showed a normal complete blood count, normal liver function tests, normal kidney function tests, and normal electrolytes. Inflammatory markers were elevated, with an erythrocyte sedimentation rate of 50 mm/hour and a C-reactive protein level of 178.6 mg/L. Venous blood gas analysis showed no significant abnormalities. Given the acute respiratory symptoms and markedly elevated inflammatory markers, a concurrent respiratory infection or pneumonia was initially considered, and empirical antibiotic treatment was started.

Chest radiography showed diffuse bilateral high-density micronodular opacities, predominantly involving the middle and lower lung zones. HRCT of the chest demonstrated widespread sand-like miliary calcified nodules and coalescent parenchymal microcalcifications throughout both lungs, with relative apical sparing. The calcifications were mainly distributed along the subpleural and peribronchovascular regions. Interlobular septal thickening produced a crazy-paving appearance, and the characteristic black pleura sign was also present (Figure 1).

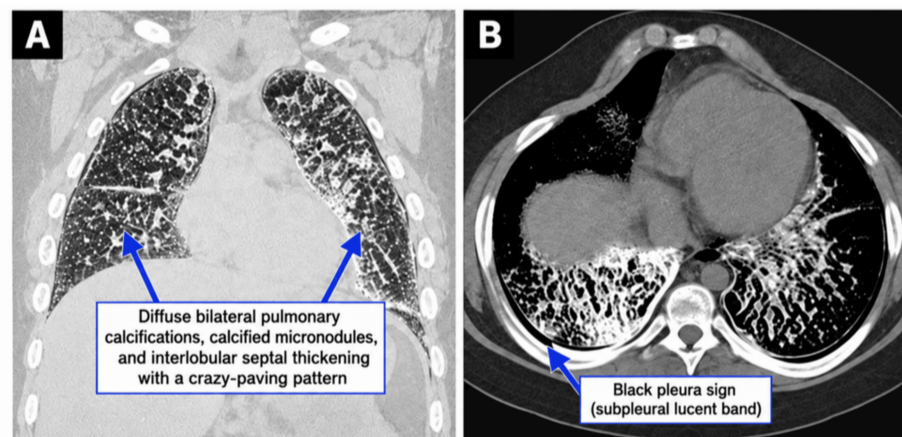


FIGURE 1: High-resolution computed tomography (HRCT) of the chest.

(A) Coronal HRCT image showing diffuse bilateral pulmonary calcifications, calcified micronodules, and interlobular septal thickening with a crazy-paving pattern. (B) Axial HRCT image showing the characteristic black pleura sign (subpleural lucent band).

These findings were highly suggestive of PAM. Flexible bronchoscopy was subsequently performed. Cytological examination of bronchoalveolar lavage fluid demonstrated characteristic microliths together with multinucleated giant cells (Figure 2).

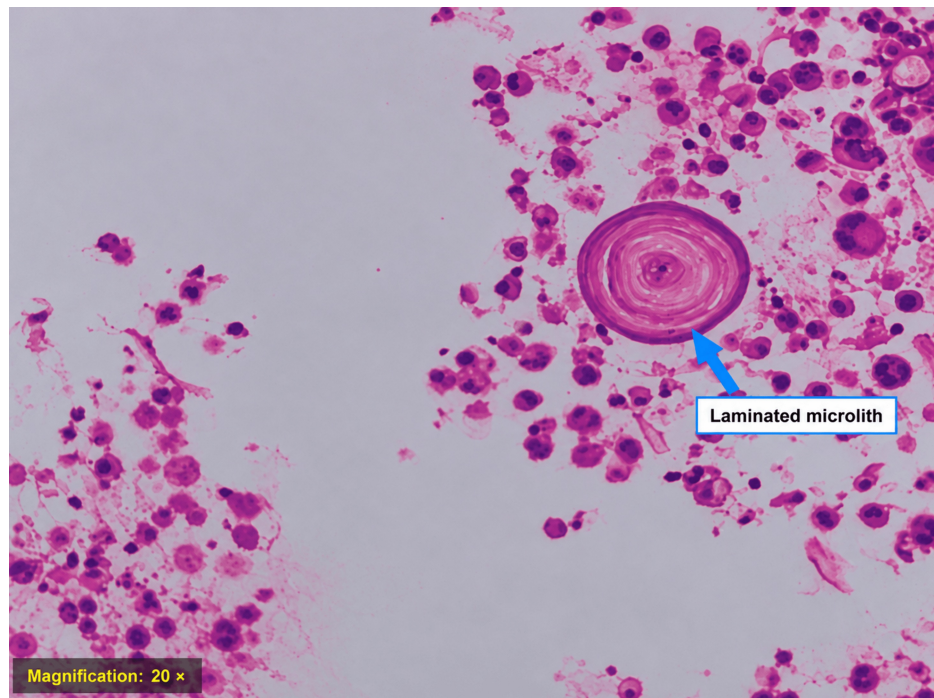


FIGURE 2: Bronchoalveolar lavage cytology (20×) showing a characteristic laminated microlith (arrow) with multinucleated giant cells in the background.

Microbiological culture yielded only normal respiratory flora. Pulmonary function testing, gas-transfer studies, exercise oxygenation measurements, and formal assessment for pulmonary hypertension were not available in the clinical record. The patient improved clinically with treatment and was discharged from the hospital.

He was subsequently followed in the outpatient clinic. Genetic analysis later identified a pathogenic variant in the *SLC34A2* gene, supporting the diagnosis of PAM. The available genetic report did not provide further details regarding the exact variant, zygosity, testing method, or classification criteria.

Following completion of the diagnostic workup, the patient was referred for lung transplantation evaluation. The detailed rationale for the referral and the subsequent outcome of the transplant assessment were not available in the accessible record. Genetic counselling was also recommended, together with screening of family members for possible *SLC34A2*-related disease.

Discussion

PAM is a rare genetic lung disorder associated with pathogenic variants in the *SLC34A2* gene [1,2]. This gene encodes a sodium phosphate cotransporter expressed mainly in type II alveolar epithelial cells. Loss of normal transporter function interferes with phosphate clearance and leads to progressive accumulation of calcium phosphate microliths within the alveoli [2].

One of the characteristic features of PAM is the marked discrepancy between the severity of radiological abnormalities and the relatively mild clinical presentation [3,4]. Our patient demonstrated this typical clinico-radiological dissociation, with extensive bilateral pulmonary calcification despite relatively mild respiratory symptoms and preserved resting oxygen saturation. His acute four-day presentation with markedly elevated inflammatory markers was considered to represent a possible concurrent respiratory infection or pneumonia; empirical antibiotic treatment was started, followed by clinical improvement and discharge.

HRCT is particularly useful in the evaluation of PAM. Typical findings include diffuse calcified micronodules, interlobular septal thickening, a crazy-paving pattern, and the black pleura sign [5]. In our patient, this combination of characteristic findings strongly suggested the diagnosis.

Bronchoalveolar lavage may provide additional diagnostic support through identification of characteristic microliths [6]. In this case, bronchoalveolar lavage cytology demonstrated microliths together with multinucleated giant cells. Genetic testing subsequently identified a pathogenic *SLC34A2* variant, providing

additional support for the diagnosis. However, the available genetic report did not provide the exact variant, zygosity, testing method, or detailed classification criteria, and the genetic result was therefore interpreted together with the concordant clinical, radiological, and cytological findings.

The diagnostic value of this case lies in the complementary use of characteristic HRCT findings, bronchoalveolar lavage cytology, and the available genetic result. In this patient, these investigations together supported the diagnosis without the need for surgical lung biopsy. This should not be interpreted to mean that all three modalities are required in every case; the diagnostic approach should be individualized according to the clinical context and the findings available.

There is currently no established medical treatment proven to stop disease progression. Corticosteroids, calcium-chelating agents, and repeated bronchoalveolar lavage have been used with limited benefit [7]. Lung transplantation remains an important treatment option for advanced disease [7,8]. Early recognition is therefore important for appropriate follow-up, supportive care, and consideration of transplantation when clinically indicated.

Recent genetic studies have expanded the known spectrum of *SLC34A2* variants in PAM. A systematic review identified an increasing number of pathogenic variants and emphasized the value of genetic testing in supporting diagnosis and recognizing familial disease [9]. More recent reviews have also highlighted the variable clinical course of PAM and the importance of family screening, particularly because affected individuals may have few or no symptoms [9]. In our patient, the available genetic result supported the diagnosis and provided a rationale for recommending screening of family members.

The clinical course of PAM can vary considerably. A recent report described a middle-aged patient presenting with advanced respiratory failure, illustrating the severe end of the disease spectrum [10]. In contrast, our patient had extensive radiological abnormalities despite relatively mild symptoms and preserved resting oxygenation. Recent reports also continue to document successful lung transplantation in patients with PAM [11,12]. In the present case, the patient was referred for lung transplantation evaluation after completion of the diagnostic workup. However, pulmonary function testing, gas-transfer studies, exercise oxygenation, formal assessment for pulmonary hypertension, and the subsequent transplant evaluation outcome were not available. Therefore, the referral itself should not be interpreted as evidence of advanced disease severity.

Overall, this case reinforces the importance of recognizing the characteristic clinico-radiological pattern of PAM and using complementary investigations to support the diagnosis while avoiding unnecessary invasive procedures. It also highlights the need for individualized follow-up, family screening where appropriate, and careful assessment of disease severity before decisions regarding advanced therapies such as lung transplantation.

Conclusions

PAM should be considered when extensive bilateral pulmonary calcification is present despite relatively mild respiratory symptoms. In this patient, characteristic HRCT findings and microliths identified on bronchoalveolar lavage cytology, together with the available *SLC34A2* genetic result, supported the diagnosis without the need for surgical lung biopsy. This case highlights the importance of recognizing the characteristic clinico-radiological pattern and using complementary diagnostic investigations according to the individual clinical context. The genetic finding was consistent with the diagnosis, while longer-term follow-up, functional respiratory assessment, and the outcome of the transplantation referral were not available.

Additional Information

Author Contributions

All authors have reviewed the final version to be published and agreed to be accountable for all aspects of the work.

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Disclosures

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