Pulmonary Alveolar Microlithiasis: a Case Report in King Hussein Medical Center (KHMC), Amman, Jordan

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ABSTRACT

Introduction: Pulmonary Alveolar Microlithiasis (PAM) is a rare disorder that can affect patients at any age, although it is more common to present in the third and fourth decades of life. Most patients are asymptomatic at the time of diagnosis. However, some may present with symptoms of dyspnea or cough. PAM can be sporadic, or it can be hereditary. Aim: To focus on the importance of using chest CT scans along with bone scintigraphy to aid in the diagnosis of PAM. The importance of screening all family members is also addressed. Case report: In our case, the patient was a 21-year-old male, coming for routine check-up to be recruited in the army. He was referred to our clinic after the examining doctor noticed that his chest X-Ray was not normal. Upon revising his chest X-Ray, he was found to have bilateral fine reticular infiltrates. His physical examination was unremarkable. His spirometry and DLCO were normal. A high-resolution chest CT scan was done, and showed diffuse bilateral microcalcifications with bilateral interstitial and septal thickening. To confirm the diagnosis of PAM, a Technetium-99m methylene diphosphonate (Tc-99m MDP) whole body bone scintigraphy was done, and it showed diffusely increased radiotracer uptake in both lungs. His family members were screened for PAM. His father and sister, who were completely asymptomatic and with normal pulmonary function tests, were found to have PAM as well. Conclusion: The use of bone scintigraphy plays an integral role in diagnosing patients with radiological findings consistent with PAM, and it can diagnose PAM without the need for invasive procedures. Once diagnosed, screening of all family members for PAM should be done, even when they are asymptomatic, as more than one-third of the cases have a familial pattern.

Keywords: Alveolar Microlithiasis, Bone scintigraphy, HRCT.

1. INTRODUCTION

Pulmonary Alveolar Microlithiasis (PAM) is a rare disease characterized by intra-alveolar deposition of calcium phosphate microliths (1). It was described for the first time by Malpighi in 1868, and was nominated by Puhr in 1933 (2). Most patients are asymptomatic at the time of diagnosis, and the diagnosis is usually suggested after detecting incidental characteristic abnormal chest X-ray or chest CT scan findings. PAM can be sporadic, or hereditary, with an autosomal recessive inheritance (3). PAM can occur at any age, however, it is more common between the third and fourth decades of life (4). There are less than 800 cases reported worldwide, with most cases being diagnosed in Asia and Europe (5). In this paper, we will focus on the importance of bone scintigraphy in diagnosing PAM in patients with characteristic abnormal chest X-ray and chest CT scan findings. Also, we will show the importance of screening all family members once PAM is diagnosed.

2. CASE REPORT

A 21-year-old male, was referred to our clinic for further investigations after being found incidentally during routine military recruitment medical check-up to have an abnormal chest X-Ray. The patient, who is a non-smoker, was completely asymptomatic at the time of presentation. He denied any previous history of chest infections or hospitalization. His physical examination was unremarkable, with normal chest examination. His routine laboratory investigations, including calcium and phosphorus levels, were all within normal limits. His chest X-Ray was reviewed (Figure 1), and it showed diffuse bilateral reticulonodular opacities. A high-resolution chest...
CT scan (HRCT) was ordered (Figure 2), and it showed bilateral intra-lobular interstitial and septal thickening and dense calcification. These radiological findings, in an otherwise asymptomatic patient, made the diagnosis of Pulmonary Alveolar Microlithiasis (PAM) highly suspected. A Technetium-99m methylene diphosphonate (Tc-99m MDP) whole body bone scintigraphy was done to prove the diagnosis (Figure 3). It showed a mild diffusely increased radiotracer uptake in both lungs. Spirometry was done for the patient, and it was normal, with a Forced Expiratory Volume in the first second (FEV1) OF 91% of predicted, and Forced Vital Capacity of 88% of predicted. His Diffusion Capacity for Carbon Monoxide (DLCO) was normal (84%). His Oxygen saturation on room air, measured by a pulse oximeter, was 98%. After the diagnosis of PAM was established in our patient, we asked to investigate his family members for the presence of PAM as well. His family consisted of his father, mother and two younger sisters (14 and 10 years old). All family members denied any respiratory symptoms. Their physical examination was unremarkable. All family members had a chest X-Ray done. Both the father and the 14-year-old sister had abnormal chest X-Rays. The father, who is a 45-year-old taxi driver, has a smoking history of 20 years. His chest X-ray (Figure 4A) showed bilateral reticulonodular infiltration, which was denser than what was seen in our patient's chest X-Ray. His HRCT (Figure 4B) showed bilateral microcalcifications with mild interstitial thickening. Again, a Technetium-99m methylene diphosphonate (Tc-99m MDP) whole-body bone scintigraphy was done to prove the diagnosis (Figure 5), and it showed a diffuse increase in radiotracer uptake in both lungs. The father’s spirometry and DLCO were normal, with oxygen saturation of 98% on room air.

The 14-year-old sister had an abnormal chest X-Ray as well (Figure 6A). Her chest X-Ray showed bilateral fine reticular infiltration, mainly in the lower lobes. Her HRCT (Figure 6B) showed microcalcifications in both lung fields. She refused to do whole-body bone scintigraphy. Her spirometry and DLCO were within normal limits, and she had an Oxygen saturation of 99% while breathing room air.

Both the mother and the 10-year-old sister had normal chest X-Rays, and they were not further investigated.

3. DISCUSSION

PAM is a rare lung disease, caused by the intra-alveolar accumulation of microliths. The diameter of these microliths ranges from 0.01 to 3 mms, and they are mainly composed of calcium phosphate (6). Many mechanisms have been suggested in the etiology of PAM, such as an inborn error of metabolism, an immune reaction to irritants, a response to some types of pulmonary insults and infections and an acquired abnormality of calcium and phosphorus metabolism (7). However, the exact etiology of the disease is still unknown. Many articles studied the role of SLC34A2 gene mutation in the pathogenesis.
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Pulmonary Alveolar Microlithiasis (PAM) is a rare lung disease. The patients are usually asymptomatic at the time of diagnosis. Typical chest X-Ray and HRCT findings can be used to diagnose PAM without the need for further investigations. Technetium-99m methylene diphosphonate (Tc-99m MDP) whole-body bone scintigraphy can aid in the diagnosis of PAM, as it may detect calcification foci in the lung parenchyma, even early in the course of the disease. In the presence of typical radiological findings, further investigations to diagnose PAM, such as bronchoalveolar lavage (BAL) and transbronchial biopsy can be avoided.

PAM usually has slow progression but may lead eventually to respiratory failure and right-sided heart failure. Although many pharmacological therapies have been studied, such as etidronate and systemic steroids, none of them showed efficacy in treating patients with PAM. At the time being, management of PAM remains supportive, with oxygen therapy and vaccinations for those who develop respiratory failure. The only curative option is lung transplantation, especially if it is performed before the disease progresses to an advanced stage. There was no documented recurrence in the patients who received lung transplantation.

In our case report, the patient was asymptomatic. Abnormal radiological findings along with an increased radiotracer uptake in both lungs by bone scintigraphy, were used to diagnose the patient. After screening his family members, his father and one of his two sisters were also found to have PAM. This shows the importance of screening family members of patients with PAM. The patient and his affected family members were advised to undergo regular medical follow up of their disease. However, they all lost to follow up to our clinic.

4. CONCLUSION

PAM is a rare lung disease. The patients are usually asymptomatic at the time of diagnosis. Typical chest X-Ray and HRCT findings can be used to diagnose PAM without the need for further investigations. Technetium-99m methylene diphosphonate (Tc-99m MDP) whole-body bone scintigraphy can aid in the diagnosis of PAM, especially in its early stages. Once diagnosed, screening of all family members is important, as more than one-third of the cases have a familial pattern.
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