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Case report A SCITECHNOL JOURNAL

17 Year Old Woman with Dyspnea Circling Back for the Diagnosis

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Study about the case

Pulmonary and critical care

Chief compliant

This is a 17 year old woman who presents with progressive shortness of breath on exertion during the last 3 months.

HISTORY OF PRESENT ILLNES

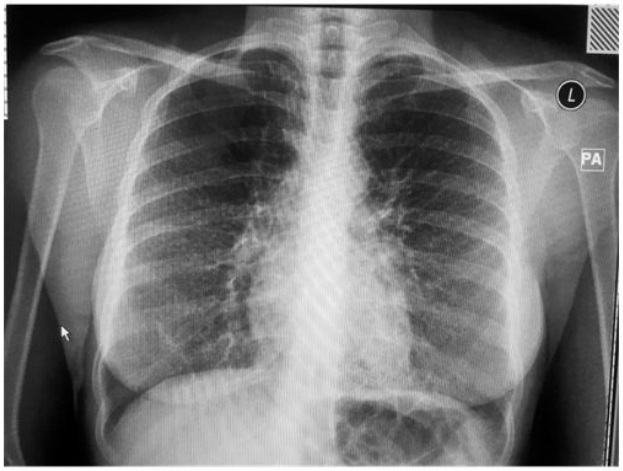
- Complains exertional shortness of breath doing sports, and climbing stairs during the last 3 months.
- The last weeks she began to feel dyspneic with moderate activity.
- 1 day ago before admission she was breathless after walking a single flight of stairs and spent most of the last 24 hours in bed.

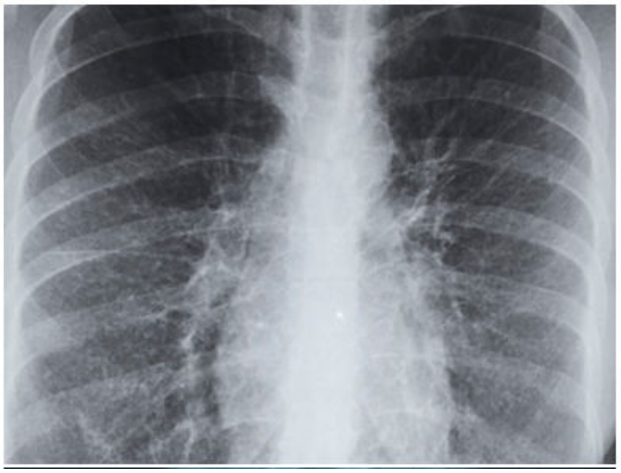
REVIEW OF SYSTEMS

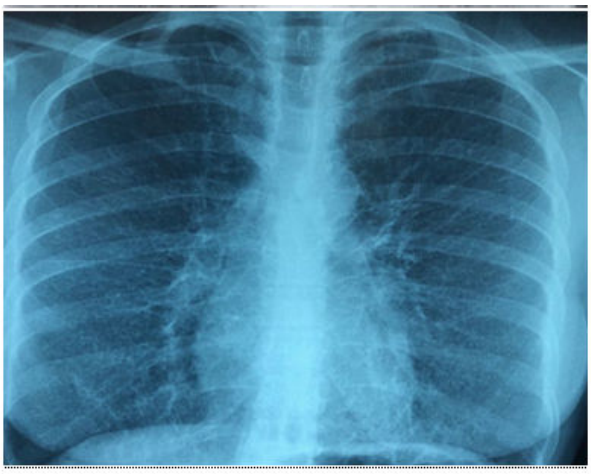
Negative for cough, chest pain, wheezing, loss of weight or any other complaints

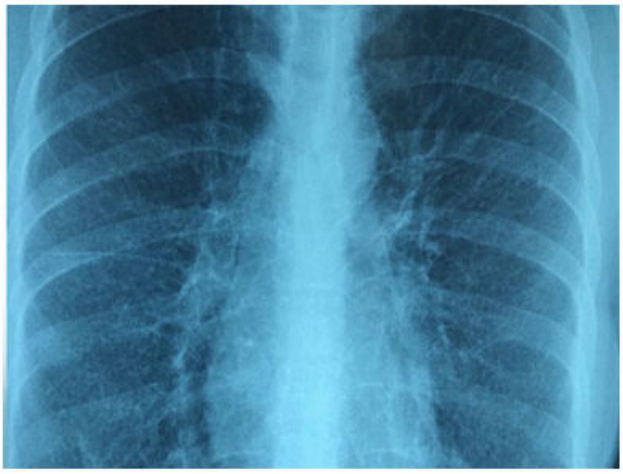
WORK UP

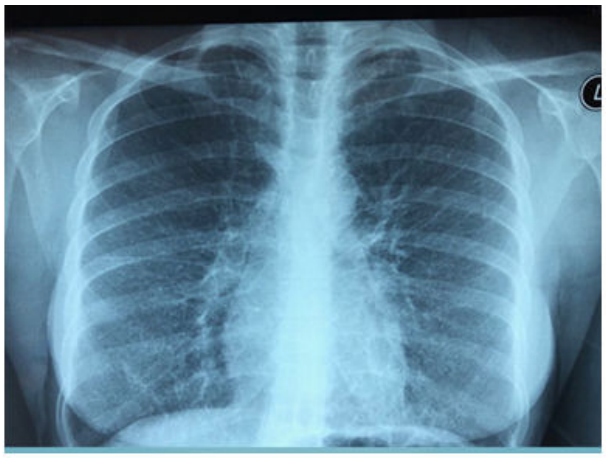
Chest x ray: revealed diffuse and bilateral reticulo- nodular (micronodular) pattern is seen, with predominance in mid and lower lobes





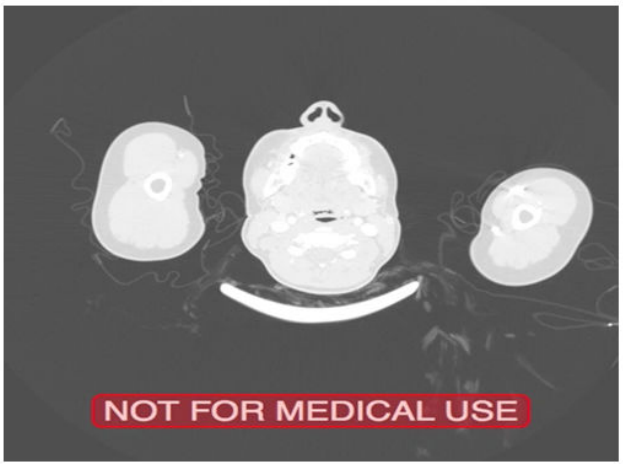


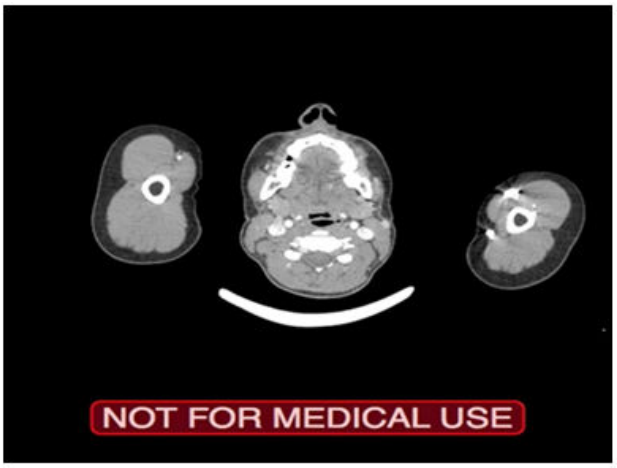




WORK UP

A computed tomographic (CT) study of the thorax after the intravenous injection of contrast material showed extensive and bilateral micro nodular infiltrates in the lower and middle lobes, with septal lines thickening.





WORK UP

- A complete blood count, and the levels of urea nitrogen, creatinine, calcium, phosphorus, magnesium, ESR, CRP, liver function, electrolytes were normal.
- The urinanalysis revealed few calcium oxalate crystals
- The arterial; blood gas while she was breathing ambient air was: PH: 7.45 mm Hg, PO2: 86 mm Hg, PCO2: 31 mm Hg, B.Exces: -1.1, HCO3: 22 mmol/L, Sat O2: 98%.

WORK UP

- PPD negative.
- ANA negative
- ACE negative
- Pulmonary function test including Lung volumes, and diffusing capacity(DLCO) were normal.
- Echocardiography: normal

WHAT IS YOUR NEXT STEP?

- Open lung biopsy
- PTH and thyroid function
- bronchoscopic exploration
- QuantiFERON - TB (IGRA)

BRONCHOSCOPY

The results of a bronchoscopic examination were negative, and cultures were negative for bacteria, including PCR for Mycobacterium tuberculosis. Cytological examination shows a smear poorly cellular and contains few endobronchial cells along with few leucocytes, without malignant cells.

Transbronchial biopsy : Revealed concentrically laminated intra-alveolar structures, no inflammatory lesions.

WHAT IS YOUR NEXT STEP ?

- start anti TB treatment and follow up after 2 months
- Start high dose IV/PO corticosteroids.
- Open lung biopsy
- ICS and follow up.

OPEN LUNG BIOPSY

The procedure was a videoassisted thoracoscopic lung biopsy. The superior segment of the right lower lobe were sampled. There was two pulmonary alveoli surrounded by concentric calcificated structures. No Inflammatory lesions were observed.

DIFFERENTIAL DIAGNOSIS

- Causes of diffuse pulmonary disease (Micronodular):
- Infectious: a) Viral: (CMV, Varicella), b) Bacterial (M. Tuberculosis), c) Fungal (H.Capsulatum, C. Neoformans).
- Inmunologic: a) Sarcoidosis, b)Extrinsic allergic alveolitis
- Neoplasic: a) Bronchioloalveolar carcinoma, b) Metastatic Carcinoma.

NOW WHAT?

- Repeat bronchoscopy
- Ask for a second opinion.
- Start corticosteroids and follow up.
- Start anti –TB treatment and follow up.

2ND READING

- Biopsies had been sent to MAYO CLINIC in USA.

- The histologic appearance revealed concentrically laminated intra-alveolar calcium phosphate microliths compatible with a Pulmonary alveolar microlithiasis (PAM) [1].

PULMONARY ALVEOLAR MICROLITHIASIS

- Pulmonary alveolar microlithiasis (PAM) is a rare autosomal recessive disease with a high penetrance characterized by widespread intra-alveolar accumulation of innumerable minute calculi called microliths.
- It is caused by inactivating mutations in the gene "solute carrier family 34 member 2", encoding a sodium-dependent phosphate cotransporter (SLC34A2).

SLC34A2 is expressed primarily in alveolar epithelial type II cells and is responsible for the uptake of phosphate released from phospholipids in outdated surfactant [2].

WORLD CASES

- Up to December 2014 a total of 1022 PAM cases were described in the literature in 65 nations.
- PAM is present in all continents, but not uniformly, the majority of cases being in Asia (576 cases; 56.3%)and in Europe (285 cases; 27.8%).
- The nation with the highest number of recorded cases is Turkey (139 cases; 13.6%), followed by China (133 cases; 13.0%), Japan (119 cases; 11.6%), India (80 cases; 7.8%), Italy (65 cases; 6.3%) and the USA (50 cases; 4.9%).
- The disease affects both sexes, with a slight predominance among males worldwide

CLINICAL COURSE

- Dyspnea on exertion occurs as the disease progresses and late symptoms of cor pulmonale appear as terminal manifestation.
- Cough may occur at any stage of disease and even the expectoration of microliths has been reported.
- Usually, the slow progression of the disease ultimately leads to fatal respiratory or cardiac failure.
- Pulmonary fibrosis, cor pulmonale and respiratory insufficiency may result with severe involvement.

RADIOLOGY

The characteristic picture of PAM on the chest radiograph shows infiltrates as fine sand-like calcific micronodules also called 'sandstorm lung', diffusely involving both lungs, usually most marked in middle and lower zones (inferior and posterior predominance), which often obliterates the mediastinal and diaphragmatic outline [3].

CO-MORBID ASSOCIATIONS

- Cases of PAM have been commonly misdiagnosed and treated as miliary tuberculosis.
- In Saudi Arabia, differential diagnosis has also been made with desert lung syndrome and non-professional pneumoconiosis due to inhalation of the desert sand.
- Other association are milk-alkali syndrome, renal transplant recipient, pericardiac cyst and lymphocytic interstitial pneumonitis.

CALCIFICATION IN EXTRAPULMONARY SITES

Calcification due to PAM has been reported involving extrapulmonary sites.

histological documentation of the presence of microliths in the lung and sympathetic ganglia and radiological evidence of microliths in gonads. Pericardial calcification and nephrolithiasis has also been reported [4].

DIAGNOSIS

- The first diagnostic clue is the characteristic chest radiograph findings. The characteristic chest radiograph and CT findings suggest the diagnosis of PAM.
- Lung biopsy (trans-bronchial or open) confirm the diagnosis. The characteristic intraalveolar lamellar microliths in the lung tissue establish the diagnosis.
- The demonstration of microliths in the bronchoalveolar lavage (BAL) fluid is possible.
- The identification of the SLC34A2 gene mutation (where available) also suggests the diagnosis.
- Serum concentrations of the surfactant proteins A and D are elevated in the patients with PAM, and can be the markers to monitor the activity and progression of the disease.

TREATMENT

- No definite treatment is available. Home oxygen therapy is necessary for the patients with respiratory insufficiency. Systemic corticosteroid and bronchoalveolar lavage have been shown to be ineffective.
- In general, no therapy has proved beneficial including whole lung lavage

Lung transplantation has been performed in a few patients. Some patients have undergone bilateral sequential lung transplantation or unilateral lung transplantations, but their long-term survival is yet to be proved [5].

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