

Photo Quiz

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TANAFFOS 

A 54 -Year-Old Man with Cough and Dyspnea

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WHAT IS YOUR DIAGNOSIS?

A 54-year-old male, farmer with a history of dyspnea from 10 years ago, developed increased sputum production, coughing and blood- streaked sputum and hospitalized for further investigations.

He was a current smoker (17pack/year) and oral opium addict. Spirometry revealed severe airflow obstruction with FVC: 45%, FEV1: 31%, and FEV1/FVC: 51%. Considering chest x-ray findings, the patient underwent chest HRCT (Figures 1 A and B). Serum immunoglobulins and IgG subclasses assessment and sweat test were performed and all were normal. Biochemistry tests were also performed, and liver and renal function tests, electrolytes, peripheral blood leukocyte counts, hemoglobin, and platelet count were all normal. ESR was 57mm/hr, and serum as well as sputum galactomannan were negative for aspergillosis. Bronchoscopy was performed; bronchoalveolar lavage fluid smear and culture were negative for pathogenic bacteria, fungi, and mycobacterium. Histopathological examination of the bronchial biopsy specimen showed chronic non- specific inflammation.

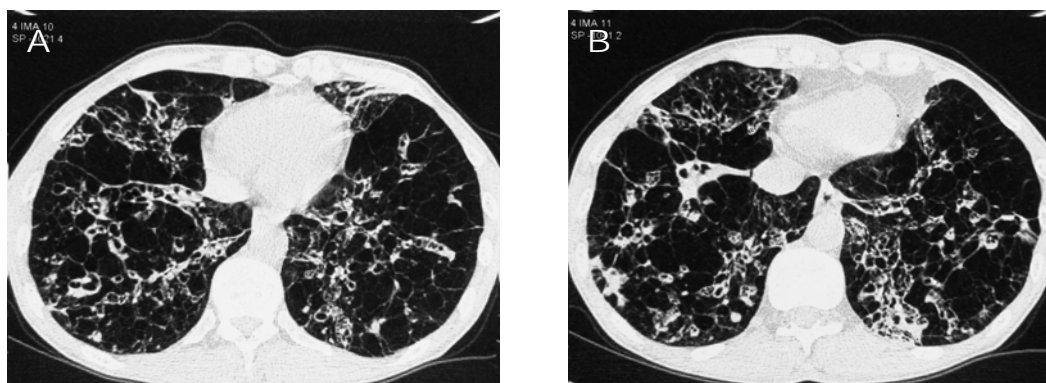


Figure 1(A,B). Chest HRCT demonstrated diffuse panacinar and cylindrical bronchiectasis in both lungs.

Answer

Diagnosis: Alpha 1-antitrypsin deficiency

Considering the chest HRCT findings which revealed diffuse panacinar emphysema and diffuse bronchiectasis, after ruling out other underlying etiologies α 1-antitrypsin serum level and activity were measured which were extremely lower than normal. Confirmatory genetic analysis detected complete mutation of gene SERPINA1. Abdominal sonography revealed cirrhotic changes in the liver.

Antibiotics and bronchodilators were prescribed and started for the patient. He was also vaccinated against pneumococcal infections and seasonal flu and underwent pulmonary rehabilitation. He was advised to quit smoking and use supplemental oxygen.

Alpha 1-antitrypsin is a protease Inhibitor that belongs to Serpin superfamily and is secreted by the hepatocytes and released into the blood stream. This enzyme inhibits neutrophil elastase and plays an important role in inflammation control and tissue repair. It is an acute phase reactant (1-2) and its deficiency occurs as the result of mutation in gene SERPINA1 located in the long arm of chromosome 14.

Risk of COPD increases in patients with α 1-antitrypsin deficiency and is mainly due to the emphysema and less frequently due to airway problems and bronchiectasis (3). Risk of COPD is higher in z protein phenotype, cigarette smokers, male gender and asthmatics. In such patients, severe early onset panacinar emphysema commonly occurs at the lower zone of the lungs but may be diffuse or with a predilection for the upper parts of the lungs (4). Other complications of α 1-antitrypsin deficiency can be neonatal hepatitis, adult cirrhosis, hepatocellular carcinoma, necrotizing panniculitis and Wegener's granulomatosis (5).

TREATMENT

Smoking cessation (if the patient is a smoker), use of bronchodilators, immunization against the seasonal flue and pneumococcal infections, oxygen therapy and pulmonary rehabilitation are recommended for all patients. Augmentation therapy (if available) is also advised for all patients with α 1-antitrypsin deficiency and fixed airflow obstruction. In the future, the new treatment methods would be inhaled form of α 1-antitrypsin, gene therapy (upon its completion and approval) and a newer technique (which is taking its first steps) for extracting the mis-folded α 1-antitrypsin from the hepatocytes into the blood stream (5).

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