Alpha-1 antitrypsin deficiency (AATD) in a young female patient

Градска општа болница
8-ми СЕПТЕМВРИ
Скопје

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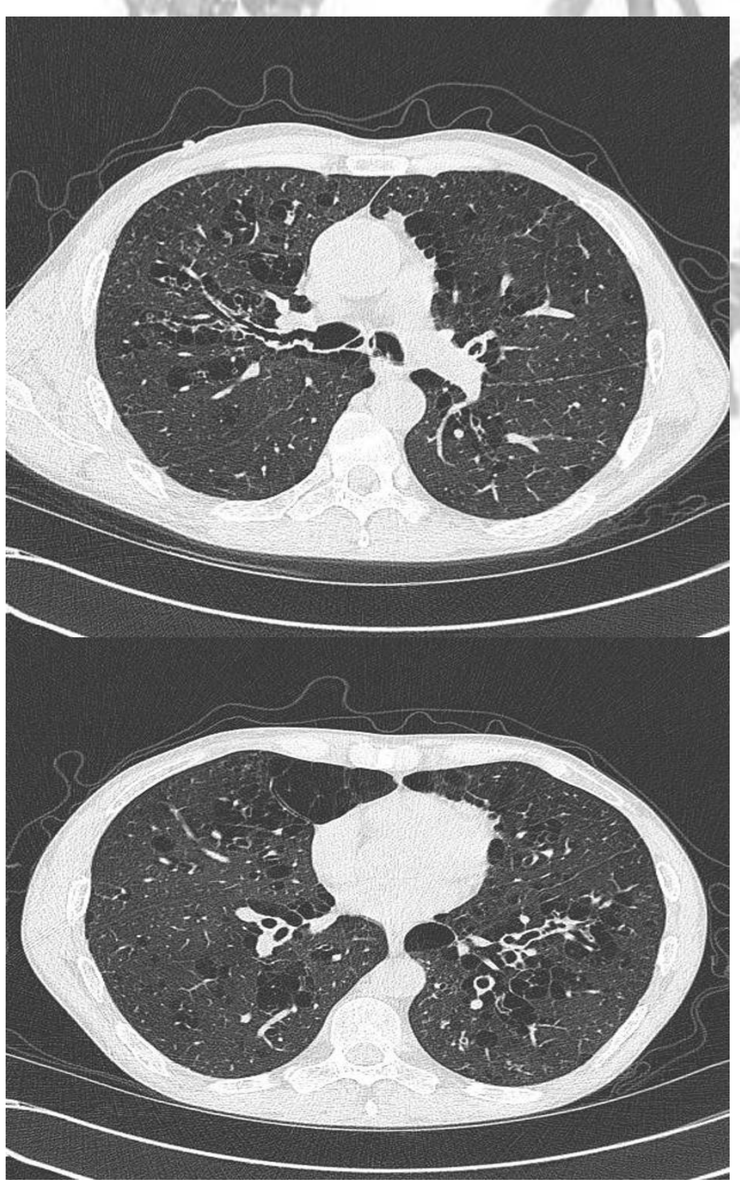
BACKGROUND

The alpha-1 antitrypsin deficiency (AATD) is a hereditary autosomal codominant disease. The phenotype Pi ZZ is associated more frequently with pulmonary disease and is responsible for the presence of emphysema early in life, particularly in smokers. Generally, AATD is suspected in young patients with pulmonary emphysema or chronic obstructive pulmonary disease (COPD). Patients often suffer from diagnostic gaps and are misdiagnosed with chronic obstructive pulmonary disease (COPD), asthma, and airway hyper-reactivity (AHR), as AATD may present with nonspecific respiratory symptoms. AATD is most common in white people, and it most frequently affects the lungs and liver. In the lungs, the most common manifestation is early-onset (patients in their 30s and 40s) pan acinar emphysema most pronounced in the lung bases. However, diffuse or upper lobe emphysema can occur, as can bronchiectasis. The most frequently described symptoms include dyspnea, wheezing and cough. Pulmonary function testing shows findings consistent with COPD; however, bronchodilator responsiveness may be seen and may be labelled as asthma.

Figure 1. Chest CT findings



CASE PRESENTATION

We describe a case of a 40-year-old Caucasian female patient, admitted to hospital because of dyspnea, malaise, cough. Symptoms started one year ago, after mild SARS-COV 2 infection. Chest X-ray during the acute illness described emphysema, with flattened diaphragm, and no signs of consolidation. According to history she was a non-smoker, office worker, with negative family history of respiratory or liver illness. She never used any regular therapy before, no comorbid diseases and denied frequent respiratory infections during childhood. Chest computer tomography (CT) presented pan acinar emphysema most pronounced in the lung bases (Fig 1). Post bronchodilator spirometry revealed forced expiratory volume in 1st second (FEV1) 54%, and forced vital capacity (FVC) 84%, with FEV1/FVC=0.64. Routine biochemistry laboratory was normal. Gas analyses noted respiratory failure type 1 (partial) with hypoxemia partial oxygen pressure 8.1kPa, hypocapnia because of hyperventilation with partial carbo dioxide pressure 4.1kPa, and oxygen saturation 92%. Echocardiography without any findings of right heart failure, normal systolic pulmonary arterial pressure (sPAP). Abdominal ultrasound without pathological findings, no liver disease detected. According to CT finding the patient was sent to the Institute for clinical immunology and genetic disorders where the serum value of alpha-1 antitrypsin was measured. The value was 0,2 micromoles /L (reference value 5-6 micromole /L). The patient was prescribed inhaled therapy of long acting anticholinergic, short acting beta-2 agonist. She was also suggested therapy with intravenous human alpha 1-proteinase inhibitor (AAT augmentation therapy).

CONCLUSION

It is never too late to suspect AATD, especially in a patient with an unusual medical history. In recent years, evidence is beginning to emerge that there may be value in identifying and treating patients who do not already have deterioration of functional parameters. The Alpha-1 Foundation recommendations for the diagnosis and management of AATD in adult patients indicate that treatment should be provided for patients with FEV_1 between 30 and 65%. It may be useful to evaluate and treat patients based on clinical symptoms, even outside the established parameters, in particular cases.

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