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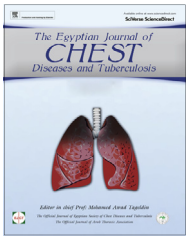

ORIGINAL ARTICLE

Alpha 1 antitrypsin deficiency in non cystic fibrosis bronchiectasis

Mohamed Shahat Badawy [a,*], Atef Farouk El Qarn [b], Hamdy Ali Mohamadeen [c]

[a] *Chest Department, South Valley University, Egypt*
[b] *Chest Department, Assiut University, Egypt*
[c] *Chest Department, Sohag University, Egypt*

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KEYWORDS

Alpha 1 antitrypsin deficiency;
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Clinical evaluation

Abstract *Setting:* It is important to identify manifestations of alpha1-antitrypsin deficiency (A1ATD) in bronchiectasis to improve care and outcome in these patients.

Objective: To clinically evaluate A1ATD in patients with non cystic fibrosis bronchiectasis.

Material and methods: Patients with non cystic fibrosis bronchiectasis were diagnosed on the basis of clinical and radiological findings. They fulfilled the inclusion criteria and divided into group (A) bronchiectasis with hyperinflation (30 cases) and group (B) bronchiectasis without hyperinflation (30 cases). All patients were subjected to history taking, pulmonary function tests, and quantitative measurements of serum A1AT by radio-immunoassay.

Results: Mean age of both groups was (50 ± 8.58) and (36.87 ± 11.35) respectively (p value 0.0001). There were significant differences in gender distribution ($p = 0.006$), and smoking history (p value 0.0001). Hemoptysis was presented in 12 cases (40%), and 20 cases (66.67%) in both groups respectively ($p = 0.04$). Dyspnea was presented in 27 cases (90%) and 19 cases (63%) for groups A and B ($p = 0.02$). There were no significant differences in sinusitis, hepatological symptoms, clubbing and family history. There were significant differences in cyanosis, edema of lower limb, chest wheeze, radiological evaluation and spirometeric parameters (p value of 0.01, 0.004, 0.001, 0.001, 0.001, respectively). Three cases (5%) of A1ATD were diagnosed among all patients one case (1.5%) in group (A) of MZ allele and two brother cases (3.5%) in group (B) of SZ allele were without statistical significance.

* Corresponding author. Tel. +20 1115454856.
E-mail addresses: mohamad_badawy@yahoo.com (M.S. Badawy), afaroukeg@yahoo.com (A.F. El Qarn), h_mohammadien@yahoo.com (H.A. Mohamadeen).

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Conclusion: A1ATD can be found in patients with bronchiectasis with or without concomitant hyperinflation. Inheritance could influence an individual's risk of A1ATD for developing bronchiectasis.

Introduction

Alpha-1-antitrypsin deficiency (AATD) is a genetic disorder characterized by low serum levels of AAT. Low plasma and alveolar concentrations in human body predispose individuals to the development of early-onset pulmonary disease, most commonly bronchiectasis, emphysema and COPD [1]. Life threatening liver disease is another possible consequence of AATD. Although it is one of the most common inherited conditions, it affects about 1 in 2000–5000 individuals – it is under diagnosed [2].

The incidence and prevalence of bronchiectasis are generally not well known and are underestimated in developing countries [3]. Although the prevalence once declined over the past years in societies with high socioeconomic status, probably due to the development of preventive medicine, especially childhood immunizations, and improvement of the living conditions and widespread use of antibiotics, nowadays bronchiectasis has been recognized more, mainly due to the frequent use of high-resolution computerized tomography (HRCT) [4,5]. Approximately 40% of patients with AAT deficiency have chronic cough and sputum expectoration [6]. The presence of bronchiectasis is recognized, but, because studies of the incidence are limited [7–10], it is recognized that further data are needed to evaluate the frequency and type of bronchiectasis, and to assess the clinical and physiologic manifestations [11].

There are isolated case reports that have suggested a putative association between bronchiectasis and AAT deficiency in the absence of emphysema [12,13]. It is well recognized that AAT deficiency is associated with the early development of emphysema, but only a limited number of studies have assessed the association between AAT deficiency and bronchiectasis. The purpose of the current study was to address and evaluate the frequency of bronchiectasis, and to assess clinical and physiologic manifestations of these patients.

Material and methods

Patients with non cystic fibrosis bronchiectasis attending to outpatient chest clinic or admitted to the Chest Department in Sohag University Hospital were enrolled in the study. The study was of one year duration and had approval from the Faculty of scientific ethics committee. Written informed consent was taken from all patients.

The patients were subdivided into two groups:

Group A: bronchiectasis with hyperinflation. (30 cases)
Group B: bronchiectasis without hyperinflation. (30 cases)

These groups of patients were diagnosed by a chest high resolution computed tomography scan.

Exclusion criteria:

1. Positive reversibility test as improvement of post bronchodilator FEV1 more than 12% on pulmonary function test,
2. Cystic fibrosis patients and infected cystic lung,
3. Past history of tuberculosis.

All patients were subjected to:

1. Comprehensive history: age, gender, smoking history, tobacco exposure
   Respiratory symptoms:
   Age of onset of symptoms
   Cough, expectoration, and dyspnea (its grade, duration)
   Frequency of acute exacerbations in last year
   Symptoms of sinusitis
   Associated diseases: hepatological, dermatological symptoms
   Family history of similar disease

2. Clinical examination
   General examination: focused on cyanosis, lower limb edema, clubbing of fingers
   Chest examination: inspection, palpation, percussion, auscultation
   Abdominal examination: focused on hepatomegaly

3. Radiological evaluation:
   chest-X-ray (postero-anterior, lateral view)
   High resolution C-T chest

4. Pulmonary function tests
   They were performed with a spirometer of computer processing (Jaeger Master Screen Diffusion, Viasys Healthcare, Gmbh, Hochberg, Germany). Age, height and weight of the subjects were entered in the spirometer. The spirometer gives two values: one is the expected value and the other is the actual value. The expected values are based on height, age and weight of the subjects. FVC, FEV1, FEV1/FVC, were measured
   Bronchodilator reversibility: The subjects received nebulized short acting B2 agonist (2.5 mg salbutamol) then the test repeated after 10–15 min (the patients did not receive bronchodilators in the past 12 h). The percentage improvement in FEV1 can be calculated as follows: (Postbronchodilator FEV1-Prebronchodilator FEV1)/prebronchodilator FEV1 X 100. A positive response to bronchodilators is an increase in FEV1 from baseline that is more than 200 ml and more than 12% of the pre-bronchodilator value
   Technique of pulmonary function tests: The actual values (FVC, FEV1, FEV1/FVC) are based on the maximal inspiration and expiration of the subjects. The pulmonary function test was conducted by seating the subject comfortably in a chair. Sterilization of the mouthpiece was done before use. The subjects were asked to perform maximum inspiration followed by maximal exhalation. Three tests were performed and the subjects were assisted to improve their efforts. The best of the three performances of FVC, FEV1, FEV1/FVC were taken [14]

5. Arterial blood gases
   ABL 700 SERIES. Arterial blood gas analysis refers to the measurement of PH and the partial pressures of oxygen (O_2), SO_2, carbon dioxide (CO_2) in arterial blood and HCO_3.
   Technique of arterial blood gases: Place the patient's hand in the supine position with the wrist extended. Identify the radial artery by palpating the pulse; choose a site where the pulse is prominent. Clean the sampling site with an alcohol wipe and wait until dry. Then insert the needle slowly. When the needle is in the artery a flash of pulsatile blood will appear and obtain at least 3 ml of blood before withdrawing the needle.