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ORIGINAL ARTICLE

Recommendations for aetiological diagnosis of bronchiectasis

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Abstract The number of bronchiectasis diagnoses has increased in the last two decades due to several factors.

Research carried out over the last years showed that an aetiological diagnosis could change the approach and treatment of a relevant percentage of patients and consequently the prognosis.

Currently, systematic investigation into aetiology, particularly of those disorders that can be subject to specific treatment, is recommended.

Given the complexity of the aetiological diagnosis, the Pulmonology Portuguese Society Bronchiectasis Study Group assembled a working group which prepared a document to guide and standardize the aetiologic investigation based on available literature and its own expertise. The goal is to facilitate the investigation, rationalize resources and improve the delivery of care, quality of life and prognosis of patients with bronchiectasis.

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Rationale

Bronchiectasis (BE) is characterized by irreversible airways dilatation and lesion associated with a vicious cycle of inflammation, recurrent infection and progressive bronchial damage.

In the last few years there have been many signs of a renewed interest in this pathology due to its prevalence which had not decreased in response to a better control of respiratory infections as would be expected. This fact is mainly due to a better diagnostic capacity, the recognition of the association between BE and some systemic pathologies and also to the increase in the population survival rates, namely chronic patients. Therefore, BE epidemiological characteristics have been changing, with less post-infectious BE, more adult patients and more cases associated with other prevalent diseases like chronic obstructive lung disease (COPD).[1,2]

BE can result from several local or systemic inherited or acquired diseases. In a significant number of cases aetiology cannot be defined, even after an extensive evaluation. Therefore, the percentage of diagnoses obtained varies widely in published series.[3–5]

Obtaining an aetiologic diagnosis is often considered unnecessary and non-cost effective.[6] However, there are several reasons why one should investigate aetiology:

- The specific treatment of certain diseases causing BE relates to a better prognosis.[3,4,7]
- The presence of BE worsens the underlying disease prognosis, accelerates loss of pulmonary function, increases mortality and significantly reduces quality of life.[8,9]
- The diagnosis of hereditary diseases is important to assess transmission risk and offer genetic counselling
- BE is a complex, heterogeneous and possibly a multifactorial entity.[10]
- Some drugs used in cystic fibrosis (CF) are ineffective, even harmful, in non-cystic fibrosis BE,[11] thus aetiology should be considered in the analysis of clinical trials because it may influence final results
- A clearly defined idiopathic BE patients group will help underlying mechanisms investigations

In conclusion, after diagnosing BE a systematic aetiologic investigation should be undertaken.

The Pulmonology Portuguese Society Bronchiectasis Study Group convened a working group which prepared a document to guide and standardize the aetiologic investigation based on available literature and its own experience. The goal is to rationalize resources and improve the delivery of care, quality of life and prognosis of patients with BE.

Aetiological investigations

A systematic investigation of the underlying aetiology undertaken according to a predetermined algorithm increases the likelihood of obtaining a diagnosis with better resource management.

In order to establish the most likely hypotheses and carry out the most effective tests, the initial assessment must be guided by a detailed clinical history and physical examination.

Clinical history

Clinical history should include onset age, presenting symptoms, clinical evolution, previously diagnosed diseases, risk exposures, infertility history, non-respiratory symptoms and family history including consanguinity data (Table 1). These questions should be asked in a systematic way as patients may not value certain clinical data either because they do not significantly compromise their quality of life or because they are not aware of their relevance for the diagnosis.

Imagiology and microbiology

The results of sputum microbiological examination as well as imaging features, in particular the location of BE (Fig. 1) are also data to be taken into consideration.

Aetiologic diagnosis

It is common to assume/that the diagnosis is post-infectious BE when symptoms begin after a serious infection. Nevertheless, there are cases where symptoms occur only years later when another predisposing factor is established (for example, some degree of immunodeficiency). On the other hand, one should ask the patient about respiratory symptoms arising before the infectious episode as they may be related to the first exacerbation of undiagnosed BE. Therefore, despite the fact that infectious diseases are still a frequent cause of BE,[3–6,10] care should be taken in establishing the diagnosis of post-infectious BE especially in the presence of upper airway symptoms, non-respiratory symptoms or other systemic diseases previously diagnosed.

If after proper investigation aetiology is still not defined, it is important to be aware of new clinical data throughout the follow up that may require a reassessment. In some systemic diseases, such as inflammatory bowel disease and rheumatoid arthritis, BE may precede its diagnosis.[12,13] In other situations, there may be an evolution over the years (for instance, a patient with IgA deficiency may progress to common variable immunodeficiency).[14]

In certain cases, where aetiologic diagnoses are considered of exclusion, investigation has to be rigorous.

It would be impossible to address investigation of all causes of BE in this document, so only those that currently benefit from specific monitoring and treatment strategies with prognostic implications have been included.

Alpha-1-antitrypsin deficiency

Definition

Alpha-1-antitrypsin (AAT) is a glycoprotein, mainly produced in the liver and functions to protect the lung against proteolytic damage being a highly effective inhibitor of neutrophil elastase.

AAT deficiency (AATD) is defined by a reduced concentration of AAT in the serum and/or identification of a defective