Caso Clínico
Clinical Case

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Amiloidose traqueobrônquica primária – A propósito de dois casos clínicos

Primary tracheobronchial amyloidosis – Two case reports

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Resumo

A amiloidose traqueobrônquica primária é uma forma de amiloidose respiratória, caracterizada pela presença de depósitos insolúveis de proteína fibrilar amilóide, ao longo da parede das vias aéreas. É uma doença pouco frequente, sendo necessário uma amostra de tecido para estabelecer o diagnóstico definitivo, com base em características histológicas específicas.

Os autores relatam dois casos clínicos de amiloidose traqueobrônquica, descrevendo os seus sintomas e os procedimentos diagnósticos e terapêuticos que foram efectuados.

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Palavras-chave: Amiloidose, traqueobrônquica.

Abstract

Primary tracheobronchial amyloidosis is a form of respiratory amyloidosis, characterised by insoluble amyloid fibril proteins deposits along the airways wall. It is an uncommon disease, requiring a tissue sample to establish the definite diagnosis based on specific pathological features.

The authors report two cases of tracheobronchial amyloidosis, describing their symptoms and the diagnostic and therapeutic procedures that were performed.

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Introduction

Amyloidosis includes a group of diseases characterised by the extracellular deposit of fibrillar protein in one or more organs. Amyloidosis can be localised or systemic, according to the anatomic involvement, and primary or secondary, in this last case if associated with chronic inflammatory or infectious diseases or neoplasms. Primary respiratory amyloidosis is uncommon and can involve the tracheobronchial tree (in a focal or diffuse way) or the pulmonary parenchyma (as multiple nodules or diffuse interstitial infiltrate).

In tracheobronchial amyloidosis, symptoms such as dyspnoea, cough, hoarseness, haemoptysis, wheezing or stridor can be present, due to bronchial obstruction.

The authors describe two cases of localised tracheobronchial amyloidosis with distinct clinical features and management approach.

Case 1

A 68 year-old female nonsmoker presented with hoarseness, stridor and progressive dyspnoea for the last four months. The patient had cleft palate and a past history of arterial hypertension and cerebrovascular disease, with no neurological sequelae. She had worked as a dressmaker, and retired 3 years ago. On physical examination, she had respiratory distress, with stridor audible over the trachea. At laryngeal observation, oedema and irregularity of glottic and infra-glottic region were recognised. Full blood count, biochemical and arterial blood gases (while breathing room air) analysis were within normal values. Chest radiography showed no abnormalities. Computed tomography of the chest showed circumferential wall thickening of the trachea and the main bronchi, with no relevant lung parenchyma abnormalities (Fig. 1). Pulmonary function tests revealed the following values (as % predicted): FVC: 101%, FEV1: 87%; FEV1/FVC: 71%; PEF: 48%; FEF 25-75: 58%; FEF 50: 53%; FEF 75: 76%; VC: 96%; TLC: 87%; RV: 86%; RV/TLC: 41%.

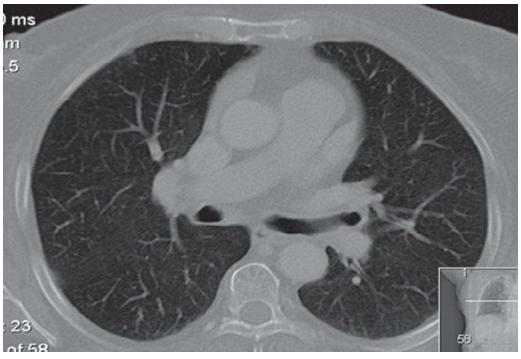


Fig. 1 – Thoracic CT scan: circumferential wall thickening of the main bronchi

Subsequently, the patient underwent fiberopticbronchoscopy, which documented, besides great deformation of ari-epiglottic folds and glottis, mucosal irregularity along the trachea and both proximal bronchial trees. There were several mucosal and submucosal yellowish lesions, some of which were flat and some were projecting from the tracheal wall to the tracheal lumen. Since two mucosal prominences, most evident in infra-glottic region, determined significant obstruction, the patient was submitted immediately to rigid bronchoscopy. During this procedure, mechanical dilatation was performed, followed by insertion of a stent (*Dumon*, 16/30 mm) in the upper tracheal portion. At this time, biopsies from the infra-glottic lesions were obtained. When she

woke up from the intervention, she no longer presented stridor, and a few hours later she was discharged from the hospital. However, four days later, the patient returned to our department complaining of acute onset of stridor and dyspnoea. Fiberopticbronchoscopy showed distal migration of the tracheal stent, causing partial obstruction in the distal third of the trachea. The stent was removed by rigid bronchoscopy. After these procedures a significant increase in the tracheal lumen was obtained and the patient totally recovered from her symptoms, and has been clinically stable since then.

Histopathological result of tracheal biopsy revealed extensive deposit of an amorphous and eosinophilic substance in the corion, with green birefringence on Congo red stain, confirming the diagnosis of amyloidosis (Fig. 2).

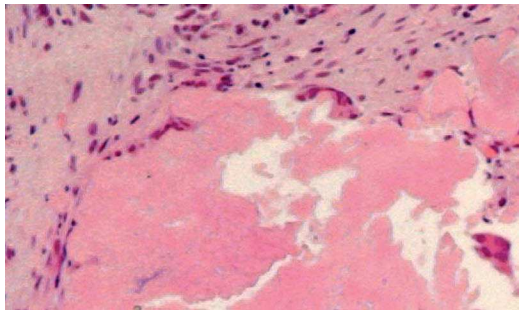


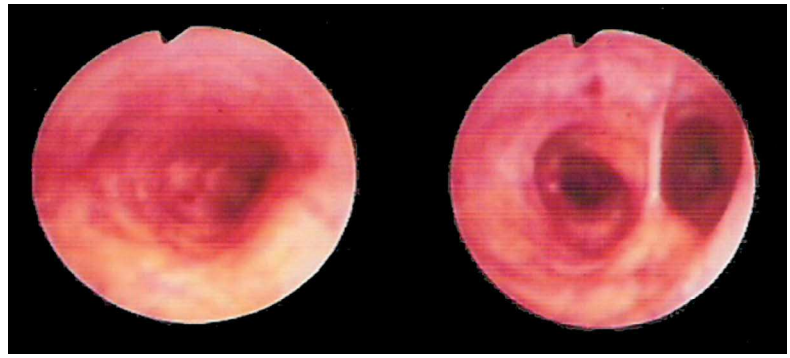
Fig. 2 – Histopathological exam: extensive deposit of an amorphous and eosinophilic substance in the corion

Presence of Bence Jones proteins in urine was excluded. Serum protein electrophoresis was normal, as well as the electrocardiogram and the echocardiogram.

From the bronchoscopy procedure until now (30 months later), the patient remained clinically well, with neither stridor nor dyspnoea and without the need of any bronchodilator or anti-inflammatory therapy. Follow-up lung function shows stability.

Case 2

A 62 year-old female nonsmoker presenting one-year history of hoarseness and dyspnoea was sent for evaluation at bronchoscopy unit for exclusion of tracheobronchial amyloidosis. She was previously observed at the Otorhinolaryngology department where indirect laryngoscopy was performed. A right ari-epiglottic fold deformation and erythema in the posterior third of the right vocal cord, with a suspicious granuloma in the posterior commissure were seen. Biopsy of these lesions confirmed the presence of extensive corion occupation by an eosinophilic, hyaline and green birefringence material to polarised light, after Congo red stain, consistent with laryngeal amyloidosis. Fiberopticbronchoscopy showed mucosal oedema with yellow-coloured plaques scattered along tracheal and main bronchi anterior wall (Fig. 3 and 4). These lesions did not cause any obstruction and were biopsied. Tracheobronchial amyloidosis has also been confirmed. Chest radiography was normal and thoracic computed tomography scan did not show any



Figs. 3 and 4 – Several mucosal and submucosal yellow-colored plaques scattered along tracheal and main bronchi anterior wall

tracheobronchial airway or pulmonary parenchyma abnormalities. Her pulmonary function test had the following values (as % predicted): FVC: 115%; FEV1: 105%; FEV1/FVC: 76%; PEF: 91%; FEF 25-75: 67%; FEF 50: 86%; FEF 75: 41%; VC: 111%; TLC: 94%; RV: 81%; RV/TLC: 35%.

Systemic amyloidosis was excluded by performing an abdominal fat biopsy, echocardiogram, serum protein electrophoresis and detection of Bence Jones proteins in urine, which were all negative.

Discussion

Amyloidosis consists in extracellular accumulation of insoluble protein, which results from the misfolding of fibril precursor proteins[1,2]. It encompasses a group of diseases, classified according to fibril precursor protein, including primary systemic amyloidosis, reactive systemic amyloidosis related with hereditary or acquired chronic inflammatory diseases (for instance, rheumatoid arthritis and chronic infections), systemic amyloidosis associated with monoclonal gammopathy or myeloma, Alzheimer's disease, type II diabetes mellitus and localised forms of amyloid deposit in single organs[3].

Amyloidosis may involve different portions of the pulmonary tract, occurring in one of three forms, namely, tracheobronchial, diffuse interstitial pulmonary infiltrates and single or multiple pulmonary nodules[3, 4, 5].

Fiberopticbronchoscopy is useful because it can reveal yellow-coloured submucosal plaques, which suggest the diagnosis and, at the same time, it allows the performance of a tissue biopsy, which is necessary to confirm the diagnosis.

The management approach of patients with tracheobronchial amyloidosis depends on their symptoms. Bronchoscopy debulking or laser resection is required in some patients with relevant airway obstruction[5,6,7,8,9]. As we described, a successful and stable result was obtained in our first patient, who underwent bronchoscopic dilatation, with mechanical resection of obstructive amyloid deposits, followed by placement of an endotracheal stent. Sometimes bronchoscopic debulking must be repeated because amyloid deposits tend to recur[9]. However, in our patient a successful removal of tracheal amyloid deposit was achieved, with normal airway patency in such a way that the stent had to be subsequently removed. On the other hand, in asymptomatic patients or in those with few symptoms and with no airway compromise, as was the case of our second patient, an observational attitude may be the best option[7].

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