Arch Bronconeumol. 2018;54(4):235–239

Sociedad Española de Neumología y Cirugía Torácica SEPAR

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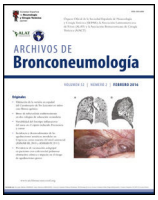

Letters to the Editor

Successful Endoscopic Resection of an Endobronchial Lipoma Using a Percutaneous Gastrostomy Snare Device

Resección endoscópica con éxito de un lipoma endobronquial utilizando un dispositivo percutáneo de asa para gastrostomía

Dear Editor:

Endobronchial lipomas are rare benign tumors which are amenable to endoscopic management both for diagnosis and treatment. Excluding those cases that merits surgical treatment, endoscopic mechanical debulking in conjunction with other techniques actually are the preferred method of treatment as it results in complete resection and preserve lung parenchyma.[1] These tumors are poorly vascularized and patients rarely present with hemoptysis, and if that is the case, it has been mainly attributed to post obstructive pneumonia or bronchiectasis rather than the tumor itself.[2] With all of this in mind, we recently decided to resect a previously histologically confirmed endobronchial pedunculated lipoma which obstructs the intermediary bronchus (Fig. 1A) from a 69-year old never smoker man who has a history of recurrent right lower pneumonia and with the aid of a cold snare device (Fig. 1B) intended for percutaneous endoscopic gastrostomy (Cook Medical). In a similar way as described by other authors using a polypectomy electric snare,[3] we grasp the endobronchial lipoma with the snare introduced in the 2.6 mm working channel of an Olympus LF-TP flexible fiberscope which in turn was placed through the rigid bronchoscope to the right main bronchus. The open snare was carefully positioned around the lipoma taking care to grasp its pedicle. Once the snare has been closed, a clean cut was obtained under the large part of the lipoma with minimal blood loss. The resected lipoma and the scarce residual lipomatous tissue were finally removed with a large forceps (Fig. 1C).

The successful use of an electrosurgical snare to resect endobronchial lipoma has already been described by other authors,[3–5] but actually there are no reports using a cold snare. Our method requires a precise diagnosis since other endobronchial lipomatous tumors that are radiologically and macroscopically very similar with lipomas are more amenable for bleeding complication.[2] Already corroborated the correct histopathological diagnosis, a second endoscopic procedure will be required to resect the lipoma. Our diagnosis of endobronchial lipoma was performed during a first rigid bronchoscopy and by means of large excisional biopsy that reasonably excluded other pathologies. During the second endoscopy, the gelatinous consistency of the tissue did not allow us to complete the removal of the large tumor remnant with only forceps. Endobronchial lipomas generally initiate in the submucosa and histological analysis of the lesions reveals lobes of mature fat cells, either partially or totally surrounded by a fibrous capsule without a prominent vasculature. This is the reason why in situations where conventional bronchoscopic ablative therapies are not available, an endobronchial neoplasia compatible with a lipoma could be resected with an improvised instrumentation with minimal risk of bleeding.

If there is a risk of long-term recurrence of this tumor using endoscopic debulking techniques, this is an issue yet to be defined by researchers, but for the moment our main goal was to avoid recurrent post-obstructive infections and their sequelae.

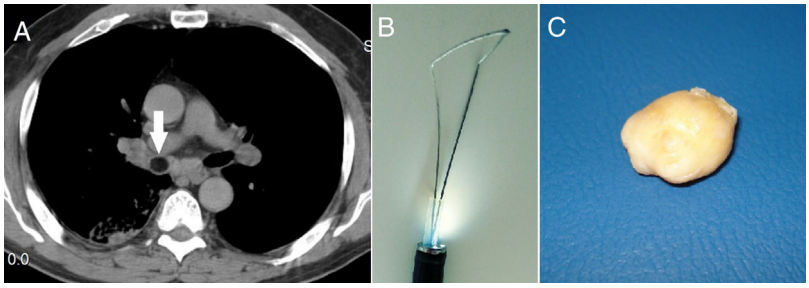

Figure 1. (A) Computed tomography showing the endobronchial lipoma blocking the right main bronchus (white arrow). (B) Our snare mounted in the flexible bronchoscope. (C) 15-mm lobulated soft tissue mass of almost excised lipoma.

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René Agustín Flores-Franco*, Luis Fernando González-Calzadillas, Stephanie Cota-Castro

Internal Medicine and Thoracic Surgery Divisions, Regional General Hospital "Dr Salvador Zubirán Anchondo", Chihuahua, Chih., Mexico

* Corresponding author.
E-mail address: rflores99@prontomail.com (R.A. Flores-Franco).

https://doi.org/10.1016/j.arbres.2017.10.004
0300-2896/

Molecular Detection of the Frequent Allele F52del in Alpha 1 Antitrypsin Deficiency

Detección molecular del alelo F52del frecuente en la deficiencia de alfa-1-antitripsina

Dear Editor,

In some recent issues, such as that of Belmonte et al.[1] published in the International Journal of COPD, have incorporated of the Mmalton allele-specific genotyping assay in the diagnostic algorithm of alpha-1 antitrypsin deficiency (AATD) to allow the clinical characterization of Mmalton individuals.

Current laboratory tests for AATD involve the determination of a combination of alpha-1 antitrypsin (AAT) serum levels, AAT phenotyping by isoelectric focusing, and an allele specific genotyping assay to detect the most prevalent, S and Z, deficiency alleles.[2] However, rare variants can only be detected by more complex techniques, such as the use of allelic specific probes or sequencing of the SERPINA1 gene, which are not available in all routine laboratories.

A study published by Martinez Bugallo et al.[3] emphasizes that the Mmalton variant in the third most frequent variant deficiency in our island, after S and Z alleles, and with a higher prevalence than that described in the Iberian Peninsula. Forty two patients with AAT values <100 mg/dL and with an inconclusive result in the genotype for PI*S and PI*Z underwent complete sequencing of the SERPINA1 gene. Of the 42 patients studied, at least one infrequent deficient allele was detected in 90.4% of the cases (38 patients). The most common deficient variant was Mmalton allele (64.2%), followed by Mpalermo allele (16.6%), both caused by the F52del mutation (Table 1).

Table 1
Genotypes of Subjects with Rare Deficiency Alleles.

Genotype	*N*	%
PI*M/Mmalton	17	40.4
PI*S/Mmalton	7	16.6
PI*Z/Mmalton	1	2.4
PI*Mmalton/Mmalton	2	4.8
PI*M/Mpalermo	6	14.3
PI*S/Mpalermo	1	2.4
PI*M/Q0amersfoort	1	2.4
PI*Z/Q0amersfoort	1	2.4
PI*Z/Q0cardiff	1	2.4
PI*MI	1	2.4
No deficitary allele	4	9.5
Overall	42	100

The Mmalton and Mpalermo are two rare variants characterized by an F52del (c.226_228delTTC) mutation. While the Mmalton allele must have derived from the normal M2 allele, Mpalermo derives from the normal M1V.[4,5]

In our population, Mpalermo represents 1 in 5 individuals with the F52del mutation, and although the use of specific probes for the detection of this mutation seems to be a good diagnostic strategy, it should be used as screening, since that in our opinion it is necessary to perform the complete sequencing of SERPINA1 in all cases to make a more accurate diagnosis of these variants, being necessary to confirm the presence of the base allele M2 or M1V in cis in these patients.

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Francisco Martínez Bugallo[a,*], Juan Marco Figueira Gonçalves[b], María Dolores Martín Martínez[a]

[a] *Human Genetic Unit, Clinical Analyses Services, University Hospital Nuestra Señora de Candelaria, Santa Cruz de Tenerife, Spain*
[b] *Pneumology and Thoracic Surgery Service, University Hospital Nuestra Señora de Candelaria, Santa Cruz de Tenerife, Spain*

* Corresponding author.
E-mail address: fmarbug@gobiernodecanarias.org
(F. Martínez Bugallo).

https://doi.org/10.1016/j.arbres.2017.10.006
0300-2896/