

Huge Para-esophageal Mass of the Mediastinum Mimicking Achalasia:

Case Report

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Abstract

Introduction: Mediastinal masses represent a difficult challenge in terms of their diagnosis and treatment. We present a case of a patient clinically mimicking as achalasia.

Case-report: A 61-year-old man, with symptoms interpreted as achalasia, presented a huge mediastinal mass suspicious for sarcoma. Only definitive pathological examination after radical surgical resection evidenced a high-grade pleomorphic sarcoma of uncertain origin.

Discussion: Mediastinum is a rare primary site of sarcoma, clinical diagnosis and pathological classification remain challenging. Surgery is still the only treatment affecting survival, even if it is not always possible. In our experience surgical radical removal was possible after a multicompartamental approach and the patient is alive with no evidence of disease after nine months from surgery.

Conclusion: Diagnosis of mediastinal sarcoma is still very difficult because of lack of pathognomonic symptoms and diagnostic work-up, clinical and pathological heterogeneity. Surgical aggressive approach is the only therapeutic option to extend survival.

Keywords: Mediastinal sarcoma; Huge mass; Rare tumor

Introduction

Mediastinal masses represent a difficult challenge in terms of their diagnosis and treatment. The real incidence is unclear in the literature due to several reasons: different classifications, inclusion of benign lesions, inclusions of systemic disease as lymphomas [1].

In this framework, soft tissues sarcomas are malignant neoplasm originating from mesenchymal tissue, arising less than 1% of malignant tumors [2]. Most frequently is diagnosed in the extremities or within the abdomen, in a prospective epidemiological study the new cases of sarcomas localized in the thorax were observed in 17%, involving lung, mediastinum, heart and pleura as main localizations [3]. Primary mediastinal sarcoma is very rare achieving about 1-9% of thoracic origin [4,5], moreover in this localization a great variety of tissue types and heterogeneity were described [6-8].

These characteristics of disease limited availability of the data, in the literature monocentric studies on a small subset of patients were published, so a lack of diagnostic work-up and treatments guidelines is noticeable. Clinical features and symptoms are not pathognomonic, diagnosis often occurs occasionally, in this report we present a case of a patient affected by mediastinal soft tissue sarcoma submitted to surgical resection clinically presenting as achalasia.

Case Presentation

A 61-year-old man, fully active and able to carry on all performance without restrictions (ECOG Performance Status 0) with a medical history of hypertension (pharmacologically treated) and a surgical history of appendectomy in the adolescence, presented at our Institution for a weight loss of about 6-7 kg since four months, dysphagia mainly for solids and regurgitation of undigested food at each meal and daily retrosternal pain. This setting of symptoms was interpreted as achalasia reaching an Eckardt score [9] of 10 as reported in Table 1. Preoperative staging included esophageal manometry, which evidenced 14 failed attempts of wet swallowing in the absence of simultaneous contractions, concluded for an achalasia of type II of the Chicago classification [9]. The preoperative esophagogastroduodenoscopy, instead, did not revealed any morphological alteration of the upper gastrointestinal tract with a normal mucosa but a defective low esophageal sphincter relaxation. Routine preoperative laboratory tests were normal. The patient was, thus, sent to surgery and a robotic Heller-Dor myotomy was programmed in September 2019. During laparoscopic peritoneal exploration, in course of operation, a modest quantity of serous fluid in the hepatorenal and paracardial space was observed and a sample was picked up and sent for a cytologic examination. During the esophagogastric junction mobilization a bulging originating above the hiatal region, strictly adherent and indissociable from the posterior face of the right diaphragmatic pillar, and an augmentation of the volume and consistence of the nodes in the station 7, 8 e 9 was detected. These findings posed the suspect of a cardia neoplasia, so another esophagogastroduodenoscopy was performed intraoperatively showing a tight stenosis at the esophagogastric junction (at about 40 cm from the upper dental arch) covered by a hyperemic mucosa in the absence of pathologic alterations that was biopsied randomly. In view of the strong suspicion of a malignant neoplasia the operation was interrupted after a surgical incisional biopsy of the mass and of the increased left gastric artery nodes. All the histologic cytologic samples examined did not reveal the presence of cancer cells. A total body and cranial CT scan for stadiation was carried out later, which confirmed the presence of the extrinsic paracardial mass of about 8 cm of maximum diameter, narrowing the distal esophagus and, subsequently, determining a dilatation of the tract above and which had a close contact with some structures such as the pancreas tail and the gastric fundus. No certain node or distant metastasis or other primitive mass were detected, even if a modest quantity of peritoneal and thoracic liquid was present. Thereafter, the echoendoscopy performed revealed a cardial stenosis not surmountable with the instrument, caused by an hypoechogenic mass of 54 x 44 mm originating from the external layers and covered by a normal mucosa which showed an inhomogeneous enhancement after the injection of intravenous contrast medium. The fine needle aspiration biopsies of the paracardial mass contextually carried out evidenced the presence of rare atypical ovoid-shaped cellular elements with a nucleolus nucleus and an immunochemistry expression profile negative for CKAE1/AE3, TTF1, CD56, S100, CD 45. The patient was finally scheduled for a thoraco-phreno-laparotomy approach and a surgical resection. The neoplasia, hourglass-shaped, passed through the diaphragm and got in touch with different structures: in the supradiaphragmatic compartment it infiltrated posteriorly the adventitial plane of the descending aorta, anteriorly it took contact with the pericardium whereas the lateral margins extended till the inferior pulmonary ligament and the inferior pulmonary vein on the left and till the right inferior lobe pleura on the right. The portion of the intraperitoneal neoplasia did not infiltrate any organ at this level.

The mass was, hence, excised “en bloc” with the lower third of the esophagus including cardia, fundus and the upper body of the stomach, a little section of the right inferior pulmonary lobe, the peri-hiatal diaphragm and its pillars. The margins extemporaneous histologic examination was negative for the research of cancer cells. The postoperative course was characterized in the third postoperative day from a right basal pleural collection treated by percutaneous drain. Liquid intake was possible at day 5, whereas solid diet was administered at day 7. Canalization to gas was referred at day 4 and at stool at day 6. The patient was, then, discharged at day ten; the CT scan with oral contrast enhancement performed at postoperative day 16 did not reveal any contrast spillage at the anastomosis level. The definitive histologic findings deposited for a high-grade pleomorphic sarcoma of uncertain origin with a cellular proliferation index (Ki-67) of 80% and a high mitotic index of 35 mitosis/10HPF which some of them atypical. The immunohistochemical phenotype analyzed was as follows: ActinSM-, Sarcomeric Actin-, MYOD1-, Desmin focally positive, pS100-, CD34-, Calretinin-, CD117-, DOG1- stressing the malignant and undifferentiated behavior. The 3, 6, 9 and 12-month-Follow-Up did not revealed any relapse of disease.

Table 1: Patient’s achalasia severity assessed by the Eckardt score.

Weight loss (kg)	6-7	2
Dysphagia	Each meal	3
Retrosternal pain	Daily	2
Regurgitation	Each meal	3
Eckardt score	Total	10

Discussion and Conclusion

Mediastinal huge masses are a clinical entity uncommon and extremely heterogeneous including both benign and malignant disease.

A standard diagnostic approach to these patients is not defined, diagnosis is very difficult because rarity of cases, lack of pathognomonic symptoms, large heterogeneity of natural history and clinical features are described.

In the field of neoplastic masses, mediastinum is a rare primary site of sarcoma, most frequently is diagnosed in the extremities or within the abdomen especially in the retroperitoneal space. In a prospective epidemiological study, the new cases of sarcomas localized in the thorax were observed in 17%, involving lung, mediastinum, heart and pleura as main localizations [3]. Due to these reasons, publications about primary mediastinal sarcoma are few out of some case-reports or small series [1,4,10], and a recent retrospective cohort analysis of data from National Cancer Database reported results about 976 patients [8].

The absence of randomized controlled trials leading decision making in the diagnosis and treatment of mediastinal sarcoma, is determined by the difficulty to enroll homogenous patients, mediastinal sarcoma including a variety of histologic tumor types, each of which grouped into rare cancers.

Furthermore, diagnosis of sarcoma is difficult even after pathological examination because of heterogeneity classification of disease between authors [4,11]. Luo et al, for instance, excluded from their evaluation the malignant peripheral nerve sheath tumor because of neurogenic origin [10], while Burt et al included them in the mediastinal sarcomas group [4]. The fine needle aspiration does not contributed to clarify criteria for diagnosis of soft tissue thoracic mass, different cytological features were observed in several subtypes of sarcomas [12]. The final pathological examinations, confirmed heterogeneity of morphological and immunohistochemical features of sarcomas and often different panel are considered in different reports [13].

It follows that a real protocol of diagnostic procedures is not described and, consequently, diagnosis of mediastinal mass is often occasional, as in our case where patient's symptoms and instrumental findings depose for achalasia. As reported in the case-report session, our patient, after specialist consultation, esophagogastroscope and esophageal manometry, was diagnosed with achalasia, scored as class III according to Chicago classification, and the gastroenterologist addressed the patient to us for minimally-invasive robotic myotomy. Intraoperative diagnosis was, as reported, challenging and reached only after complete removal and pathological analysis. Finally, according to the 2013 WHO classification [14], the histological findings were compatible with high-grade pleomorphic sarcoma.

On this basis, the extemporaneousness of diagnosis is cause of trouble in the staging of disease above all when great vessels and nearby structures are involved, consequently the therapeutic approach to the mediastinal masses is not codified [2,4,8,10].

The role of surgical resection is clear from prior studies on survival [3,7,8], utility of other treatments is debated, but surgical treatment is possible in less than half of the patients and of whom just one third is radically resected [6], in particular in mediastinal sarcomas which are often huge or invading at the moment of diagnosis and the location is close to vital organ. Recently, a review showed how an aggressive surgical approach even in case of involvement of great vessels could remain the only therapeutic option and salvage surgery is superior to medical palliation [15].

In our experience surgical removal was possible after a multicompartimental approach to expose mediastinal space, mass and all the vital organs included in the anatomic spaces involved.

Nevertheless, radical surgery was possible, postoperative course was satisfactory and the patient is alive with no evidence of disease after nine months from surgery.

In conclusion diagnosis of mediastinal sarcoma is very difficult, lacks of mediastinal pathognomonic signs, limited contribution of radiological images, heterogeneity of tumors and scarce incidence are the main reasons. The work-up for diagnosis and staging of mediastinal sarcoma are not edited, consequently guidelines to treatment are not validated, surgical aggressive approach is the only therapeutic option to extend survival.

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