A rare case of extraskeletal osteosarcoma of the esophagus: an example of difficult diagnosis

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SUMMARY: A rare case of extraskeletal osteosarcoma of the esophagus: an example of difficult diagnosis.

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Sarcomatous lesions of the esophagus are rare. We describe a controversial case of a malignant aggressive tumor of the esophagus, with a very poor prognosis and rapid outcome for the patient.

A 74-year-old man underwent endoscopic examination for recurrent thoracic pain and dysphagia. A 8 cm mass was found in the cervical esophagus. A sarcomatous tumor with osteoid aspects was observed on the histopathological examination, without any carcinomatous component.

KEY WORDS: Extraskeletal osteosarcoma - Esophageus - Rare tumors - Conservative treatment.

Introduction

Extraskeletal osteosarcomas (EOS) are rare and malignant neoplasms characterized by the production of osteoid matrix. They represent 1–2% of all soft tissue sarcomas and 4% of all osteosarcomas (1). They share histomorphologic features with osteosarcomas arising in the bone, but are not related to bony structures (2). All major subtypes of osteosarcoma that arise in bone can also be found in EOS. The most common is the osteoblastic variant, with abundant osteoid, followed by fibroblastic, chondroid, telangiectatic, small cell, and well-differentiated types (2). The limbs is the most common anatomic site (47%), followed by the upper extremity (20%) and retroperitoneum (17%) (2-5). The literature includes cases of EOS arising in unusual sites, such as the larynx, tongue, small intestine, colon, liver, gallbladder, heart, urinary bladder, parotid, pleura, lung, mesentery, diaphragm and breast (1-3, 11). To our knowledge only one case of esophageal EOS has been reported (12).

Most of the malignant primary tumors of the esophagus are of epithelial origin. Sarcomas of the esophagus are very uncommon (1% to 1.5% of all esophageal tumors) (13). They are basically divided into two types:
- pure sarcomas of mesenchimal origin, usually leiomyosarcoma (14, 15), less commonly liposarcoma (16, 17), malignant fibrous histiocytoma (18) and Ewing’s sarcoma (19);
- tumors with mixed pattern of epithelial and spindle cell characteristic, such as synovial sarcoma (20, 21) and carcinosarcoma (12, 13, 22-24).

The term “carcinosarcoma” refers to a rare biphasic neoplasm comprising squamous carcinoma and sarcomatous cells. The carcinomatous component is a squamous cell carcinoma, but rare cases of adenocarcinoma have also been described. The sarcomatous component is usually made up of spindle sarcomatous cells resembling a malignant fibrous histiocytoma, but it sometimes shows a differentiation towards muscle, cartilage or bone (12, 13, 22-24).
We describe the rare case of primary osteosarcoma of the esophagus and its difficult and controversial differential diagnosis (23).

Case report

A 74-year-old man in a poor clinical status underwent esophagogastroduodenoscopy for recurrent thoracic pain and dysphagia. A large polypoid 8 cm mass was observed in the cervical esophagus. The tumour showed firm consistency and whitish colour. Four minute fragments and two major macrobiopsy of 1.5 cm each were taken.

The microscopic finding was a very pleomorphic and high grade malignant tumor (Fig. 1). We observed cellular areas, with sheets of anaplastic epithelioid or spindle cells arranged in solid fashion, with intermixed necrotic areas. These cells had vesiculous and sometimes nucleolated nuclei, presenting large cytoplasms with focal clear aspects. Mitotic rate was high, with abundant atypical mitotic figures. We observed foci of osteoid differentiation, showing osteoid immature matrix (Fig. 2) and focal condroid aspects with multinucleated osteoblast-like giant elements. The luminal side of the macrobiopsy showed granulation tissue, with entrapped rare squamous elements, that we interpreted as residual cells of the esophageal epithelium (Fig. 3). On the basis of routine histological stains, a diagnosis of malignant sarcomatous lesion was supposed.

For clinical reasons, the patient couldn’t be operated. In few weeks the lesion became obstructive and developed haemorrhagic complications. After two months the patient died for infective and cardiovascular disorders. It was not possible to perform radiological specific investigations to stage the disease.

Materials and methods. Two paraffin blocks of the endoscopic fragments were obtained. They were cut at 3 micron and stained by hematoxylin-eosin. On the supposed sarcomatous nature of the tumor, some immunohistochemical stained were performed on 2 micron paraffin sections by Bond Max Menarini Diagnostics immunostainer. We employed the following panel of antibodies: monoclonal smooth muscle actin (clone alfa sm-1, Menarini), monoclonal muscle specific actin (clone HHF35, Novocastra), monoclonal CD34 (clone QBEnd-10, Dako), polyclonal CD117 (Dako), monoclonal Pancytokeratin (clone 5D3 and LP34, Novocastra), monoclonal Desmin (clone D33, Dako), monoclonal Vimentin (clone V9, Menarini).

Results

We observed the sarcomatous anaplastic cells on immunohistochemical slides. These malignant elements presented strong positivity for vimentin and mild but diffuse positivity for muscle specific actin, mild and focal positivity for smooth muscle actin and desmin. S100 protein, CD34 and CD117 were negative, such as pankeratin, that marked only epithelial squamous esophageal cells entrapped in the tumor on its luminal side (Fig. 4).
These epithelial elements were regular and didn’t show any sort of atypia. No differences on immunophenotypic expression were observed in spindle cells or in anaplastic giant-cell elements.

Discussion

We described an aggressive malignant sarcomatous lesion without evidence of epithelial malignant component. It consisted in an intraparietal tumoral mass ulcerating the overlying mucosal squamous epithelium. The most peculiar histologic picture of this neoplasia was the presence of osteoid differentiation with immature condroid aspects, that induced us to conclude the case as sarcomatous malignant neoplasia with osteosarcomatous aspects.

Extraskeletal osteosarcoma are exceedingly rare mesenchymal lesions. Aesophageal osteosarcomatous tumors have been described in dogs, but only one case has been reported in humans (12, 25). Due to the rarity of the tumour, differential diagnosis can be difficult, requiring the collaboration of pathologist, surgeon and radiologist.

In our case, radiological study and surgical operation were not feasible in relation to the bad conditions of the patient and his difficult care management. The only diagnostic material has been obtained from endoscopic examination. The histologic aspect of the tumor suggested dedifferentiated anaplastic sarcomatous lesion. In the differential diagnosis we considered extraskeletal osteosarcoma, other malignant mesenchymal tumors and carcinosarcoma with osteosarcomatous differentiation and osteoid component. Immunoprofile of the tumor can be concordant with other as malignant as infrequent sarcomatous lesions, such rhabdomyosarcoma or malignant histiocytoma, but in these cases we couldn’t explain the osteoid malignant component as a malignant trasformation of a metaplastic osseous event. The presence of osteoid structure and the absence of epithelial malignant cells (pankeratine negativity) were consistent with osteosarcoma.

These rare and aggressive digestive tumors are interesting, especially when they affect patients without past clinical history, in absence of genetic involvement. Some sporadic somatic genetic mutations could be supposed to be responsible for extraosseous osteosarcoma (2). To date there is few evidence about the genetics of EOS, for the rarity of the lesions but also for the extremely aggressive evolution.

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References

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