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Sporadic perivascular epithelioid cell tumor of the wall of the jejunum: A case report

BACKGROUND: *Perivascular epithelioid cell tumors are rare mesenchymal tumors composed of histologically and immunohistochemically distinctive perivascular epithelioid cells. They are considered ubiquitous tumors and have been described in different organs, but gastro-intestinal PEComas are diseases of extreme rarity.*
METHODS: *We report a case of a 51-year-old woman, without a medical history of tuberous sclerosis complex, affected by abdominal PEComa, adhering tightly to the jejunal loop and to the spleen.*
RESULTS: *During the surgical operation, a large abdominal mass was found, and surgical resection was carried out. Definitive histologic and ultrastructural findings were consistent with PEComa.*
CONCLUSION: *Given the rarity of GI-PEComas and the lack of cases reported in the literature, we want to emphasize the importance of conducting further studies in this regard, to better describe their biological behaviour.*

KEY WORDS: Gastro-intestinal tumor, PEComa, Perivascular epithelioid cell tumors

Introduction

Perivascular epithelioid cell tumors (PEComas) were first termed by Bonetti et al. in 1992 [1,2]. The World Health Organization has defined them as "rare mesenchymal tumors composed of histologically and immunohistochemically distinctive perivascular epithelioid cells (PECs)" [3]. PECs usually express melanocytic and myogenic markers such as actin, desmin, calponin, human melanin black (HMB) 45, melanA, and microphthalmia-associated transcription factor (MITF) [4].
Under the definition of PEComas are included angiomyolipoma, lymphangioleiomyoma, clear-cell myomelanocytic tumor of the falciform ligament, clear-cell "sugar" tumor of the lung and other locations [5]. Apart from these mentioned subtypes, PEComa-not otherwise specified (PEComa-NOS) are also reported; they have been described in different organs and are considered ubiquitous tumors.
To our knowledge, gastro-intestinal (GI) PEComa-NOS is a disease of extreme rarity: fewer than 40 cases have been published to date, and the sigmoid colon was the most frequent location [2,3].
Here we report a rare case of sporadic PEComa arising in the wall of the jejunum in a 51-year-woman.

Case Report

A 51-year-old Italian woman was admitted to our hospital because of gradual onset of abdominal pain and heaviness and nausea. In 1992, the patient had undergone sigmoid resection for diverticulitis. Then, she had

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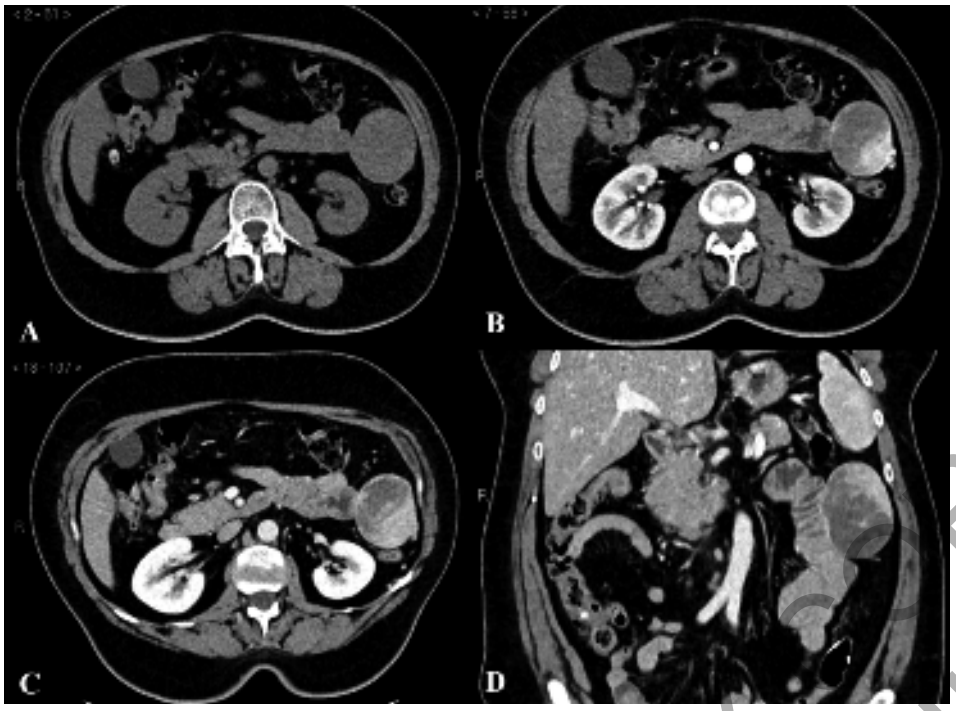

Fig. 1: Intravenous contrast-enhanced CT revealed a rounded, lobulated mass, measuring 6x5.8x8cm (APxLLxCC), adherent to the wall of the first jejunal loop, hypodense on basal acquisition (A) and, on contrast enhanced acquisition, showing peripheral enhancement with central hypovascularity (B, arterial phase. C, portal phase. D, coronal plain).

Fig. 2: Microscopic findings.
Hematoxylin and eosin staining, 10x magnification (A) and 20x magnification (B), showed epithelioid and spindle cells with abundant clear to granular eosinophilic cytoplasm. No mitotic figures were identified. The pseudocapsulated aspect is visible (B).

undergone right quadrantectomy for breast cancer in 2015, with subsequent radiotherapy and chemotherapy. In 2017 she also had undergone left quadrantectomy. The patient did not have a medical history of tuberous sclerosis complex. Her family history was unremarkable. Physical examination revealed a diffuse pain on palpation of the upper quadrants. All blood and biochemical tests were within the normal ranges. An intravenous contrast-enhanced whole body computed tomography (CT) scan revealed, between the leaflets of the mesentery in the left hypochondrium, a rounded mass, with lobulated borders, measuring 6x5.8x8cm (APxLLxCC), adherent to the wall of the first jejunal loop, hypodense on basal acquisition and, on contrast enhanced acquisition, showing peripheral enhancement with central hypovascularity (Fig. 1). Because of her medical history, the patient had many previous TC examinations. In particular, the last one, performed in 2017, did not show any abdominal lesions, therefore it seemed that the mass has risen and grown in recent months.

She was scheduled for elective surgery. During the surgical operation, a large abdominal mass was found, which measured about 10 cm in diameter, adhering tightly to the jejunal loop and to the spleen. After lysis of adhesions and ligation of the vascular pedicle, surgical resection of the tumor was carried out.

On definitive histology, the morphology of the cells was fairly uniform (Fig. 2); the epithelioid cells had abundant eosinophilic, at times granular, cytoplasm, PAS-positive. The nuclei were large, with vesicular nucleoli. No mitotic figures were identified. These cells were arranged in nests, divided by sheets of connective tissue. These histologic and ultrastructural findings were consistent with those of PEComa, and immunohistochemical features further confirmed the diagnosis. The tumor had a pseudocapsulated aspect, and this could be considered an indicator of benignity.

The patient was alive and well with no signs of recurrence or metastasis for 6 months of follow-up.

Discussion

Here we report a case of a sporadic abdominal PEComa adhering to the wall of the jejunum. PEComas are able to originate in nearly every organ all over the body. These tumors are found most often in the kidney and in the uterus, while in the GI tract are rare. Among the GI tract, the most favourite site of the lesion is the colon (45.7%) followed by the small intestine (28.6%), rectum, stomach, and esophagus [5]. Additionally, the sigmoid colon seems to be the most frequent location among different portions of the colon [3]. These tumors are commonly associated with tuberous sclerosis, although they can also be sporadical [2].

The clinical presentation is usually not specific. Depending on their location and size, they could be counted among the rare causes of intestinal obstruction, bleeding, non-specific abdominal pain, or symptoms of compression of the surrounding structures [3,6–9]. Ultrasound, CT and magnetic resonance are the imaging tests used, but the definitive diagnosis is immunohistological.

Histologically, these tumors are composed of epithelioid and spindle cells with abundant clear to granular eosinophilic cytoplasm. They are typically arranged around blood vessels in characteristically formed nests or sheets, often infiltrating the smooth muscle of small- to medium-sized vessels [10]. Immunohistochemically, the tumor cells are positive for melanocytic markers, including HBM-45 and Melan A, and in most cases are positive for myocytic markers, including smooth muscle (actin and desmin) markers [2,5,11].

The differential diagnosis includes gastrointestinal stromal tumors (GIST), smooth muscle tumors, metastatic melanoma, and endocrine tumors. PEComa can mimic GIST when it has a spindle cell morphology or expresses CD117. Also the morphology of leiomyomas or leiomyosarcomas can be very similar to that of PEComas. Only immunohistochemical analysis can provide a definitive diagnosis of PEComa [7].

Surgery is currently the treatment of choice for primary tumors, local recurrence, and metastases [9,12,13].

Since PEComas are extremely rare tumors, it is often difficult to describe their biological behaviour. Although clear criteria for malignant PEComa are yet to be established, most recently it has been reported that large tumor size, infiltrative growth pattern, high nuclear grade, and high proliferative activity are possible predictors of aggressive behavior [2].

Conclusion

PEComas are rare mesenchymal tumors; they have been described in different organs and are considered ubiquitous tumors. GI PEComas are diseases of extreme rarity, and it is often difficult to describe their biological behaviour. We reported this case to implement the present literature and to emphasize the importance of conducting further studies in this regard.

Riassunto

I tumori perivascolari a cellule epiteliodi (PEComa) sono definiti come "rare neoplasie mesenchimali composte da cellule con particolari e definite caratteristiche istologiche ed immunoistochimiche". Questi tumori sono considerati ubiquitari, sebbene la localizzazione gastrointestinale sia rara: sembrano essere meno di 40 i casi attualmente descritti in letteratura. Nell'ambito del tratto gastrointestinale, la localizzazione più frequente è il colon sigmoideo.

Nel nostro lavoro, descriviamo il caso di una paziente di 51 anni di età, giunta alla nostra attenzione per un dolore addominale aspecifico e diffuso, senso di pesantezza e nausea persistente. La TC total body con mezzo di contrasto evidenziava a livello addominale, tra i foglietti mesenteriali, una massa tondeggiante, lobulata, di dimensioni 6x5.8x8cm (APxLLxCC), in continuità con la parete della prima ansa digiunale; la massa appariva vascolarizzata perifericamente dopo acquisizione di mezzo di contrasto.

La paziente è stata sottoposta ad intervento chirurgico in regime di elezione, durante il quale è stata confermata la presenza di tale voluminosa massa intra-addominale, strettamente adesa all'ansa digiunale. Eseguita la resezione del tumore, le caratteristiche istologiche e l'analisi immunoistochimica sono risultate compatibili con la diagnosi di PEComa.

Il PEComa può essere sporadico, come in questo caso, oppure essere associato a Sclerosi Tuberosa. Istologicamente è caratterizzato da cellule epitelioidi e fusate con abbondante citoplasma eosinofilo granulare, tipicamente organizzate in nidi o lamine intorno ai vasi di piccolo e medio calibro. La chirurgia è attualmente il trattamento di scelta. Essendo tumori piuttosto rari, è ancora oggi difficile definirne il comportamento biologico.

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