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Case Report

A Gastric Perivascular Epithelioid Cell Tumor: A Case Report

Baptiste Abbar[1]*, Frédéric Charlotte[1], Jean Michel Coindre[2] and Jean Philippe Spano[3]

[1]*Department of Pathology, University Pierre and Marie Curie APHP (Paris VI), France*
[2]*Department of Pathology, University Bordeaux 2, France*
[3]*Department of Medical Oncology, University Pierre and Marie Curie APHP (Paris VI), INSERM_UMRS1136, France*

***Corresponding author**

Abbar Baptiste, Department of Pathology, University Pierre and Marie Curie (Paris VI), 14 rue de buci, 75006, Paris, France, Email: baptiste.abbar@gmail.com

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Abstract

Perivascular epithelioid cell tumors (PEComas) of gastro intestinal tract are very rare, with only few cases reported in the literature. The PEComas are a family of rare mesenchymal tumors characterized by perivascular epithelioid cell differentiation. We herein report our experience with a patient who had a primary gastric PEComa that was diagnosed by an expert pathologist review of the excised tissues and which showed neopastic epithelioid cells arranged as sheets with abundant eosinophilic cytoplasm, nuclei with a prominent nucleolus and expressed melanocytic and muscle markers.

INTRODUCTION

Perivascular epithelioid cell tumors (PEComas) are mesenchymal tumors composed of distinctive cells that show a focal association with blood vessel walls and usually express melanocytic such as HMB-45 or melan-A, and smooth-muscle-actin markers. This family includes angiomyolipoma, lymphangioleiomyomatosis, clear cell sugar tumor of lung and a group of rare, morphologically and immunophenotypically similar lesions in others locations, usually called PEComas not otherwise specified.

PEComas have been reported in various anatomic sites, but most often arise in the retroperitoneum, abdominopelvic region, uterus, and gastro intestinal tract. To our knowledge, this is the fifth case of primary gastric PEComa reported and the second one arising in the antrum [1].

CASE REPORT

A 33-year-old male was admitted to the emergency department of a Montenegrin hospital for severe abdominal pain at the beginning of June 2015. The patient did not have any particular medical history and his family history was unremarkable. Neither alcohol nor tobacco use was noted. An abdominal and pelvic computed tomography scan found a 133 x 73mm heterogeneous abdominal solid mass associated with massive peritoneal effusion. Emergency surgery was performed by median abdominal laparotomy. The examination of the abdominal cavity revealed a hemoperitoneum and a hemorrhagic mass 10-cm in diameter located within the front wall of gastric antrum, four centimeters far from the Pylorus.

A partial gastrectomy with two centimeters of surgical margin was made associated with a partial omentectomy. The resection was considered macroscopically complete.

The first histopathologic reading yielded a diagnosis of dedifferentiated liposarcoma with areas of myxoïde liposarcoma and rhabdomyosarcoma.

The patient presented to our department (La Pitié-Salpêtrière Hospital Paris France) requesting for a second opinion at the beginning of July 2015. The pathologic specimen was reviewed by our institution pathologist (FC) and an expert in sarcoma pathology (JMC), coordinator of a national network for pathology review of all cases of newly diagnosed sarcoma in France. The tumor infiltrated the gastric wall and the omentum. The neoplastic cells were arranged as sheets (Figure 1A). Large areas of necrosis were observed. The neoplastic cells were epithelioid and were characterized by abundant lightly eosinophilic cytoplasm. The nuclei had a prominent nucleolus. There were three mitoses for 10 high power fields. A mild inflammatory infiltrate made up of small lymphocytes and eosinophil cells was noted (Figure 1B). The immunostaining revealed that the tumor diffusely expressed caldesmon (Figure 1C) and phospho-S6. HMB45 and Melan A markers were focally expressed (Figure 1D). Rare tumor cells were positive for TFE3. Cyclin D1 was expressed in 20% of neoplastic cells. The tumor was negative for AE1/AE3 cytokeratin, CD117, DOG1, CD34, smooth muscle actin, ALK1, transgelin and

myogenin. Ki-67 showed a proliferation rate of 25%. Given these pathologic findings, a final diagnosis of PEComa was yielded.

The reevaluation few weeks after surgery (at the end of July 2015) with a positron emission tomography computed scan and a thoracic, abdominal and pelvic computed tomography did not find any sign of residual disease. The case was discussed in our multidisciplinary tumor board, which concluded for a simple follow-up every three months for the first two years.

DISCUSSION

We reported here a case of a primary gastric PEComa treated surgically. The diagnostic criteria of PEComa-not otherwise specified are now well-established, based on tumor with a perivascular epithelioid cell differentiation which distinctively co-express melanocytic markers and muscle markers. The gastro intestinal tract is the second most common site of PEComas-not otherwise specified cases and the stomach the fourth most common site of gastro intestinal PEComas after the colon, the small intestine and the rectum [1]. Our knowledge about the genetic alterations in PEComas is still very limited.

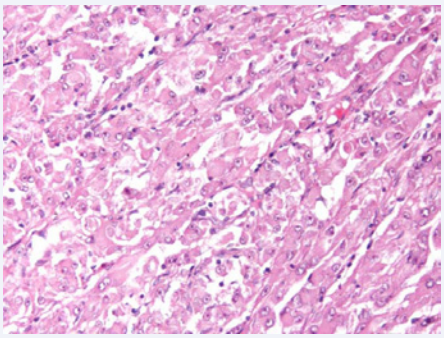

Figure 1a The tumor is composed of sheets of large epithelioid cells which harbors nuclei with a prominent nucleolus (HES, original magnification ×200).

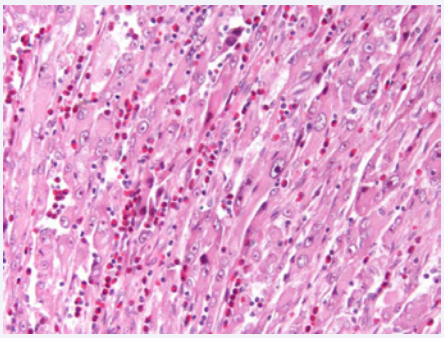

Figure 1b In this field, the tumor cells are admixed with eosinophils (HES, original magnification ×200).

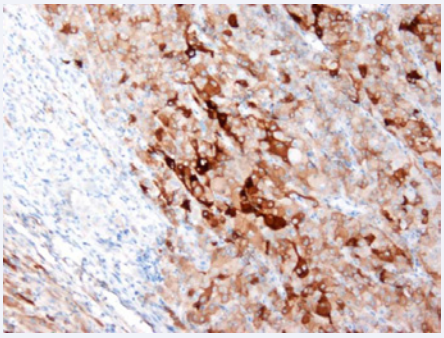

Figure 1c Diffuse expression of Caldesmon by tumor cells (Immunostaining, original magnification ×200).

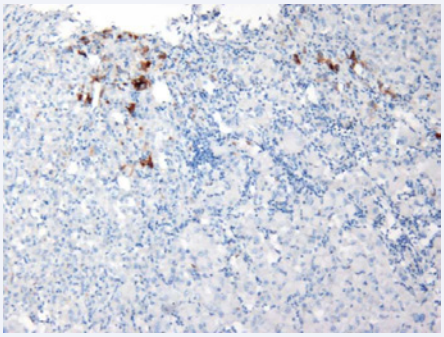

Figure 1d Focal expression of HMB45 by tumor cells (Immunostaining, original magnification ×100).

Lymphangioleiomyomatosis, clear cell sugar tumor of lung and angiomyolipoma are strongly associated with tuberous sclerosis complex but only few cases of PEComa-not otherwise specified previously reported were associated with tuberous sclerosis complex. The effect of mutations in *TSC1/TSC2* is the activation of the mTor signaling pathway. So there is a scientific rationale to target this pathway therapeutically, and it seems that mTor inhibitors are associated with a good radiological responses in tumors with genetic evidence of alterations in *TSC1* and *TSC2* [2] but also in the tumors in which no specific mutations in mTOR-related genes was found [3].

TFE3 gene rearrangements have been reported in few cases of PEComas-NOS [4]. TFE3-rearranged PEComas seem to show similar morphologic findings of a distinctively epithelioid nested neoplasm with low mitotic activity and spanned a broad spectrum of locations [4]. Those tumors may represent a distinct sub-group of PEComas and based on the prior case reports the mTor inhibitors may not be as effective in this genetic sub-group. MET inhibitors could be explored in TFE3-rearranged PEComas such as it has being done in other TFE3-overexpressed sarcomas [4].

Cyclin D1 over expression has been found in 5 cases of malignant or potentially malignant PEComas-NOS [1]. Cyclin D1 role in the pathogenesis of those tumors is unknown and may become an area of interest. The diagnosis of PEComa NOS remains difficult because of their rarity and their wide range of morphologic appearances. This case underlines the importance to subject a case of sarcoma diagnosed by a first reader to an expert review.

At the present time, based on the several cases report, surgery seems to be the most effective treatment for local PEComas of the gastro intestinal tract, and mTor inhibitors based chemotherapy for unresectable or metastatic ones, but no clinical trial was performed to test this assumption. A phase 2 clinical

trial is undergoing to confirm the efficacy of an mTor inhibitor based chemotherapy in metastatic or advanced Pecomas (NCT01690871).

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REFERENCES

1. Lu B, Wang C, Zhang J, Kuiper RP, Song M, Zhang X, et al. Perivascular Epithelioid Cell Tumor of Gastrointestinal Tract: Case Report and Review of the Literature. Medicine. 2015; 94.
2. Wagner AJ, Malinowska-Kolodziej I, Morgan JA, Qin W, Fletcher CD, Vena N, et al. Clinical activity of mTOR inhibition with Sirolimus in malignant perivascular epithelioid cell tumors: Targeting the pathogenic activation of mTORC1 in tumors. J Clin Oncol. 2010; 28: 835-840.
3. Weeber F, Koudijs MJ, Hoogstraat M, Besselink NJ, VAN Lieshout S, Nijman IJ, et al. Effective Therapeutic Intervention and Comprehensive Genetic Analysis of mTOR Signaling in PEComa: A Case Report. Anticancer Res. 2015; 35: 3399-3403.
4. Agaram NP, Sung YS, Zhang L, Chen CL, Chen HW, Singer S, et al. Dichotomy of genetic abnormalities in PEComas with the therapeutic implications. Am J Surg Pathol 2015; 39: 813-825.

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