Malignant gastrointestinal PEComa misdiagnosed as Solid pseudopapillary neoplasm of ovary: a case report

Yan Xu
Xiang Yang No.1 People's Hospital
Binghu Lin (lbhgcwk@sina.com)
Xiang Yang No.1 People's Hospital
Junping Lei
Xiang Yang No.1 People's Hospital
Song Shang
Xiang Yang No.1 People's Hospital
Jia Fu
Xiang Yang No.1 People's Hospital

Case Report

Keywords: Gastrointestinal tract, PEComa, Malignant tumor, Solid pseudopapillary neoplasm of ovary, Misdiagnosed, case report,

Posted Date: May 16th, 2022

DOI: https://doi.org/10.21203/rs.3.rs-1635516/v1

Abstract

Background: Perivascular epithelial cell malignant tumor (PEComa) is a mesenchymal tumor with the characteristics of perivascular epithelioid cells in histology and immunophenotype. It mostly occurs in kidney, uterus, liver and so on. Solid pseudopapillary is a tumor that commonly originates in the pancreas. Solid pseudopapillary of ovary is similar to PEComa in histology and is easily misdiagnosed in clinic. This paper reports a case of malignant gastrointestinal PEComa misdiagnosed as solid pseudopapillary neoplasm of ovary.

Case summary: A 35-year-old female patient came to our hospital with intermittent right lower abdominal pain for more than 1 month. After the completion of relevant examinations, the possibility of sigmoid colon tumor infiltration in the right adnexa was considered before surgery. On February 12, 2018, the tumor was surgically removed, the pathological examination showed that there was solid pseudopapillary neoplasm of the ovary. The patient received four cycles of chemotherapy with paclitaxel and platinum after surgery. The patient recovered well after operation, and there were no complications. Two years later, a pulmonary nodule was found in the reexamination, and the pulmonary lesions were surgically removed. Immunotherapy (Tislelizumab) and targeted therapy (Apatinib) are then given. After consulting several laboratories and testing institutions, the final diagnosis was (sigmoid colon) Malignant gastrointestinal PEComa involves (right) ovary and (right upper lung) lung metastasis. The patient died a month ago from complications.

Discussion: Clinicians must understand PEComa in order to achieve early detection, early diagnosis and early treatment.

Background

In 1992, Bonetti et al.[1] were the first team to put forward the concept of PEComa; In 1996, Zamboni et al.[2] found that there was pathological tissue characterized by perivascular epithelioid cells in the pancreas, which was a rare tumor from mesenchymal origin. Since then, PEComa has been paid more and more attention. In 2002, the World Health Organization (who) officially defined it as "a kind of mesenchymal tumors composed of histologically and immunohistochemically distinctive perivascular epithelioid cells". PEComa mainly includes angiomyolipoma (AML), clear cell sugar tumor of the lung (CCST), lymphangioleiomyomatosis (LAM) and some other tumors with some similarities in histology and immunophenotype[3]; Solid pseudopapillary neoplasm is a common tumor originating from the pancreas. It was first found that it originated from the ovary in the study of irving J.A et al.[4]; Solid pseudopapillary neoplasm of ovary is similar to PEComa in histology, which is easy to be misdiagnosed. This paper reports a case of malignant gastrointestinal PEComa misdiagnosed as solid pseudopapillary neoplasm of ovary.

Case Presentation

A 35-year-old female patient was admitted to our hospital on February 6, 2018 due to intermittent right lower abdominal pain for 1 month. She occasionally had swelling discomfort, increased stool frequency. Preoperative colonoscopy (Fig. 1): There are cauliflower-like new organisms 10-14cm from the rectum to the anus. Five tissues were taken for biopsy. There is a mucosal bulge about 0.3 * 0.4cm in the rectum with smooth surface.

Preoperative magnetic resonance (Fig. 2 and 3): The wall of sigmoid colon is thickened, the lumen is narrow, and soft tissue tumor can be seen in the intestinal cavity. The boundary between the soft tissue tumor and the right accessory is unclear. Preoperative chest CT showed that there were no obvious substantive and interstitial lesions in both lungs.

After completing relevant inspection, the possibility of sigmoid tumor infiltration in the right accessory should be considered before operation; On February 12, 2018, the operation was performed under general anesthesia. During the operation, the tumor was located in the right pelvic cavity. The appendix is discontinuous, the right appendix and the upper segment of rectum have formed a dense mass, and transparent mucus can be seen in the tumor; After the operation, the pathological examination in our hospital (Fig. a) showed that the tumor cells were arranged in solid nests, glandular tubes and papillae. The cytoplasm of tumor cells is transparent or eosinophilic. The nuclear groove can be seen occasionally in the nucleus which is round, The cell nucleolus is obvious, and the image of nuclear division is occasionally seen. Considering that ovarian tumors involved the appendix. The oncology department of our hospital had doubts after discussion, and sent the pathological samples to Shanghai Ackerman Medical Laboratory for consultation. After examining the pathological slides, it was considered as solid pseudopapillary neoplasm of ovary (Fig. B and C); The patient recovered well after the operation, and there were no surgical complications. The treatment regimen was 4 cycles of paclitaxel and cisplatin. Apulmonary nodule were found during reexamination of pulmonary CT in January 2020 (Fig. 4). The pulmonary nodule was considered to be malignant and was surgically removed it.

The pathological specimens were sent to Tongji Hospital for consultation. Consultation opinions: Ovarian, lung and sigmoid colon have epithelioid malignant tumors, which are considered to be Xp11 translocation mesenchymal tumors (Fig. D and E); One month later, the samples were sent to Peking Union Medical College Hospital. After consultation, it was finally diagnosed as malignant gastrointestinal PEComa of sigmoid colon involving right ovary and transferred to the right upper lung. (Fig. F and G). A year ago, the patient was receiving treatment, and the treatment scheme is adjusted to immunotherapy (Tislelizumab) combined with targeted therapy (Apatinib). She was rechecked regularly and did not complain of special discomfort symptoms.

Discussion And Conclusions

PEComa is a rare mesenchymal tumor. According to the existing relevant reports, it is mostly found in many organs such as kidney, lung and uterus. Solid pseudopapillary neoplasm are rare, with the majority of described cases originating in the pancreas[5].PEComa rarely occurs in the gastrointestinal tract, and

most of them are benign or low malignant tumors, with less distant metastasis; Gastrointestinal PEComa is usually not accompanied by characteristic clinical symptoms. Discomfort symptoms such as abdominal mass, bloody stool, abdominal pain and intestinal obstruction can appear according to the size and location of the tumor. Sometimes patients will not have any symptoms, which can be found in routine physical examination; Before surgical resection, PEComa can't be diagnosed only by clinical manifestations and imaging examination, because PEComa has no specificity in both. On CT, the tumor only shows clear boundary, uneven or uniform enhancement[6]. On magnetic resonance imaging (MRI), the lesions show bright on T1-weighted images and dark on fat- suppressed images[7]. Angiomyolipoma (AML) is the most common PEComa type. AML is categorized into two types: classic AML and epithelioid AML. Epithelioid AML is more common than classic AML in abdominal tumors or lung malignant metastases[8];

In the pathological examination, the PEComa tumor cells are arranged in a sheet or nest shape. There is a significant fibrous vascular network between the cell nests, and some tumor cells are arranged radially around the hyaline vascular wall, which has diagnostic value[9]; In the study of He, S. et al.[10], the pathological manifestations of solid pseudopapillary neoplasm of ovary are as follows: The tumor is composed of sheets and nests of cells that appear to be polygonal, interrupted by a delicate fibrous septa and capillary network, and focally by broad hyalinized bands. Therefore, the pathological examination is not specific in distinguishing PEComa from solid pseudopapillary neoplasm of ovary. In immunohistochemical detection, PEComa is mainly expressed as the co-expression of melanocytic (HMB-45, Melan-A) and smooth muscle (actin, desmin) markers to avoid misdiagnosis as leiomyosarcoma and other lesions[11]; The immunohistochemical manifestations of this patient were EMA, HMB45, TFE-3, CyclinD1,β- Catenin was positive; The diagnostic criteria of malignant PEComa recommended in the study of Folpe et al[3].It must meet the following two or more criteria: tumor size□5 cm, infiltrative growth pattern, high nuclear grade, necrosis, and mitotic activity□1/50 HPF and subsequent aggressive clinical behavior.

At present, surgical resection of diseased tumors is the first choice for PEComa. However, incomplete tumor resection may cause local recurrence or accelerate distant metastasis. For some patients with local recurrence and distant metastasis, it is necessary to choose chemotherapy as adjuvant treatment after operation{Wejman, 2015 #8}[12]. The patients reported in this paper were given four cycles of chemotherapy with paclitaxel and platinum after first surgery.And a treatment regimen after second surgery, which is immunotherapy (Tislelizumab) combined with targeted therapy (Apatinib). But now the patient has died of complications.

In general, malignant gastrointestinal PEComa is a rare tumor. There is no obvious specificity in clinical manifestations andpathological examination, and it is very easy to be misdiagnosed as other diseases with similar manifestations. Therefore, immunohistochemical detection plays an important role in the diagnosis of PEComa; The final diagnosis of this patient is mainly based on immunohistochemical detection, in which the expression of HMB-45 and TFE-3 plays a key role.Although the above two

diseases are rare and less primary in the gastrointestinal tract, they should be carefully distinguished to avoid misdiagnosis.

Abbreviations

AML: angiomyolipoma ;

CCST: clear cell sugar tumor of the lung;

LAM: lymphangioleiomyomatosis

Declarations

Ethics approval and consent to participate:
Not applicable.

Consent for publication :
Written informed consent was obtained from the patient.

Availability of data and materials:
Not applicable.

Competing interests:
None.

Funding:
The Scientific and Technological Project of Xiangyang City of Hubei Province 2021YL24

Innovative Research Program of Xiangyang No.1 People's Hospital XYY2021Q10

Contributions:
Name: Xu Yan; Lei Junping : Contributions: writing of the article / identification of papers for inclusion / performed data and evidence collection/performed the surgery.

Name: Lin Binghu; Contributions: performed the surgery

Name: Shang Song; Contributions: provide the funding

Name: Fu Jia Contributions: collect the data

Acknowledgement
Not applicable

References

1. Bonetti, F., et al., *PEC and sugar*. Am J Surg Pathol, 1992. **16**(3): p. 307–8.
2. Zamboni, G., et al., *Clear cell "sugar" tumor of the pancreas. A novel member of the family of lesions characterized by the presence of perivascular epithelioid cells*. Am J Surg Pathol, 1996. **20**(6): p. 722–30.
3. Folpe, A.L., et al., *Perivascular epithelioid cell neoplasms of soft tissue and gynecologic origin: a clinicopathologic study of 26 cases and review of the literature*. Am J Surg Pathol, 2005. **29**(12): p. 1558–75.
4. Irving, J.A. and R.H. Young, *Granulosa cell tumors of the ovary with a pseudopapillary pattern: a study of 14 cases of an unusual morphologic variant emphasizing their distinction from transitional cell neoplasms and other papillary ovarian tumors*. Am J Surg Pathol, 2008. **32**(4): p. 581–6.
5. Nguyen, M.T., et al., *Ovarian Solid Pseudopapillary Tumor Resembling Benign Hemorrhagic Cyst on Rapid Frozen Section.* Case Rep Obstet Gynecol, 2020. **2020**: p. 6473630.
6. Yeon, H.J., et al., *PEComa in the rectum: A case report and review of the literature on epithelioid angiomyolipoma*. Int J Surg Case Rep, 2021. **86**: p. 106301.
7. Chen, Z., et al., *A systematic review: perivascular epithelioid cell tumor of gastrointestinal tract*. Medicine (Baltimore), 2016. **95**(28): p. e3890.
8. Kanazawa, A., et al., *Perivascular epithelioid cell tumor of the rectum: report of a case and review of the literature*. World J Surg Oncol, 2014. **12**: p. 12.
9. Yang, Q.Y., et al., *[Perivascular epithelioid cell tumor (PEComa) of intestine: a clinicopathological analysis of three cases]*. Zhonghua Bing Li Xue Za Zhi, 2021. **50**(2): p. 128–130.
10. He, S., et al., *Solid pseudopapillary tumor: an invasive case report of primary ovarian origin and review of the literature*. Int J Clin Exp Pathol, 2015. **8**(7): p. 8645–9.
11. Dashraath, P., et al., *Malignant perivascular epithelioid cell tumor (PEComa) of the uterus*. Ultrasound Obstet Gynecol, 2021.
12. Wejman, J., et al., *PEComa of the mesentery coexisting with colon cancer: a case report*. Diagn Pathol, 2015. **10**: p. 31.

Figures

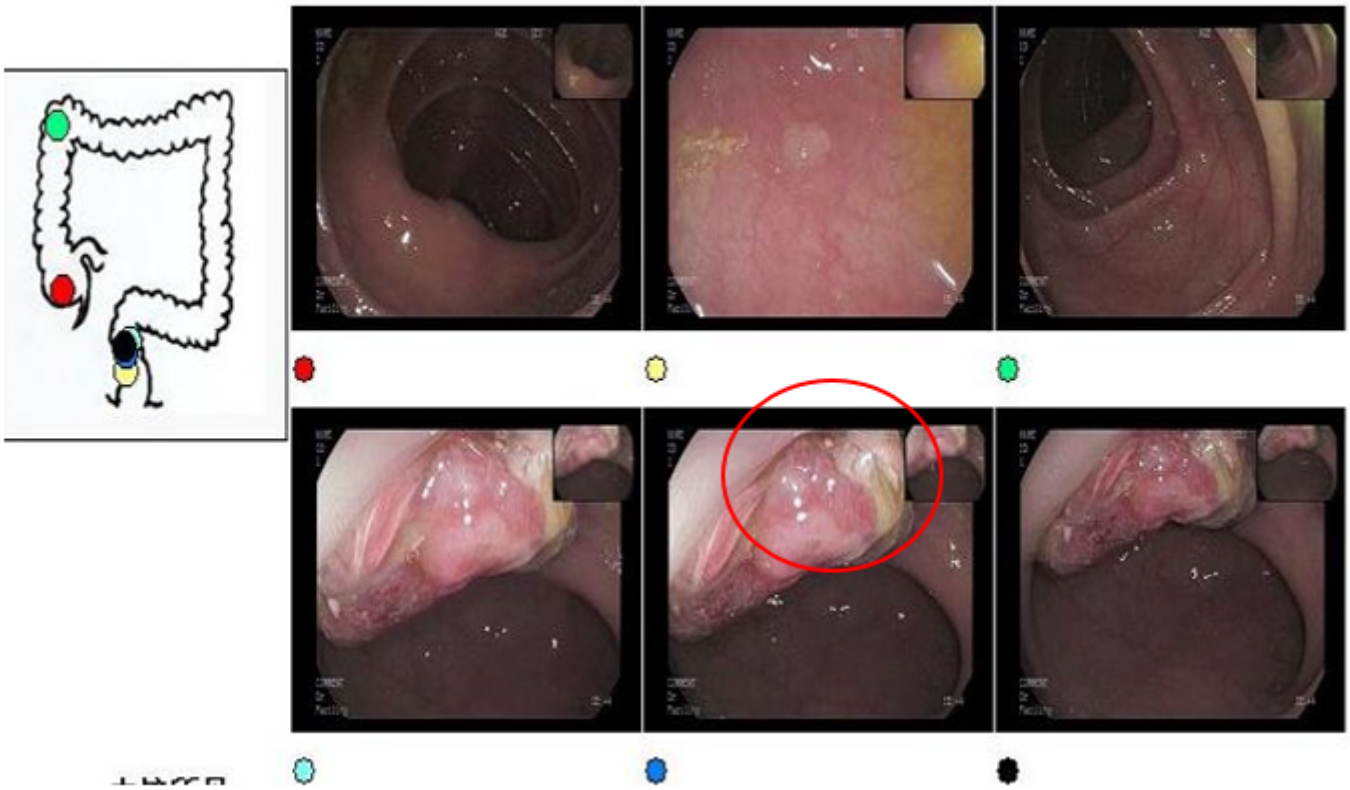

Figure 1

Colonoscopy image

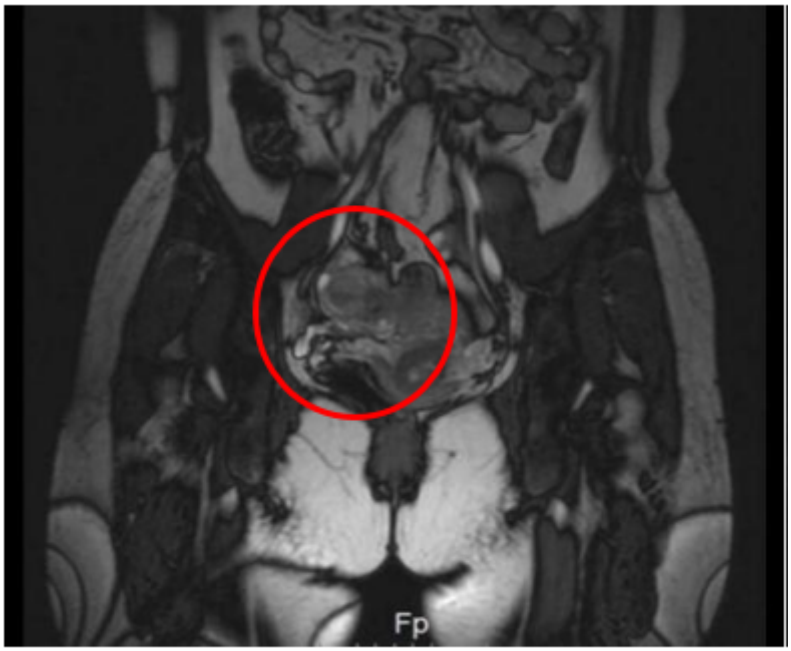

Figure 2

Preoperative magnetic resonance

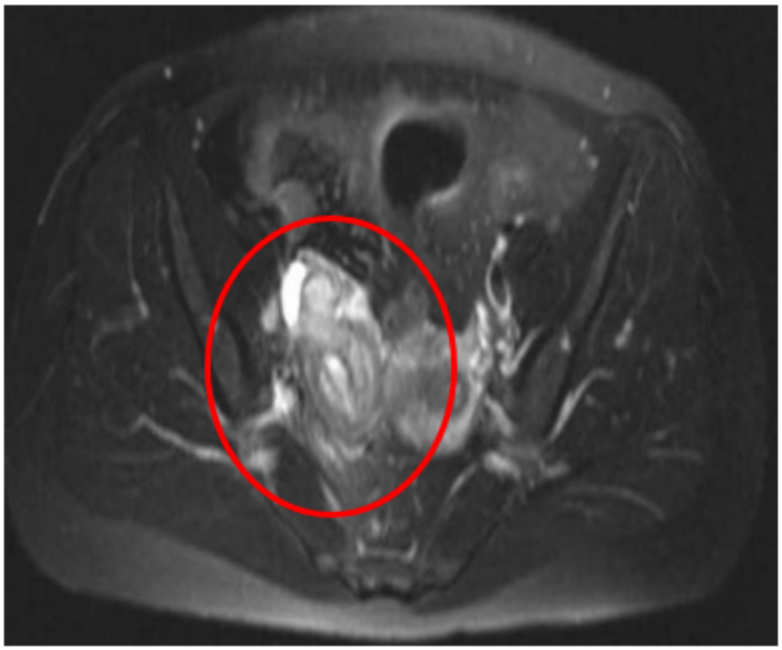

Figure 3

Preoperative magnetic resonance

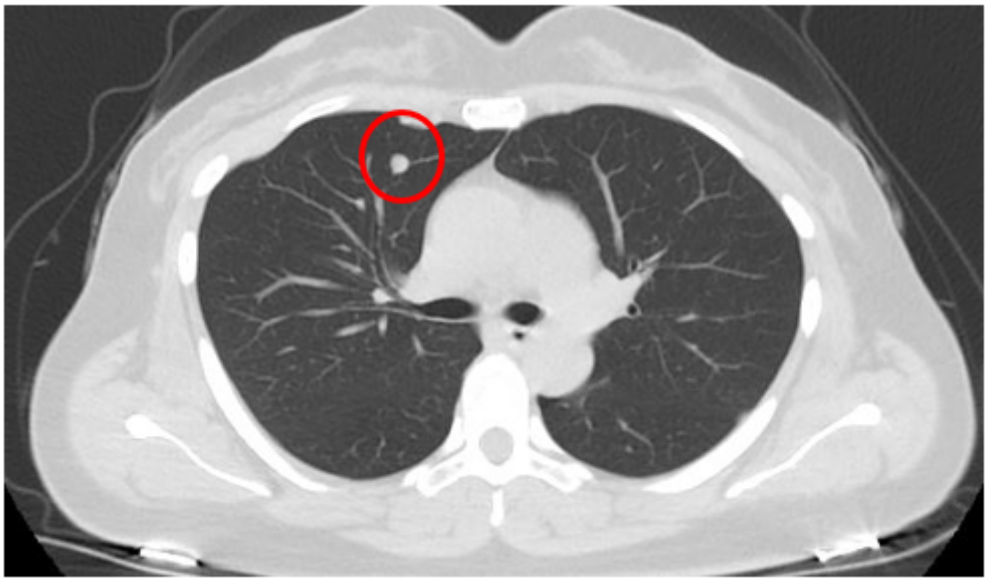

Figure 4

the pulmonary nodule

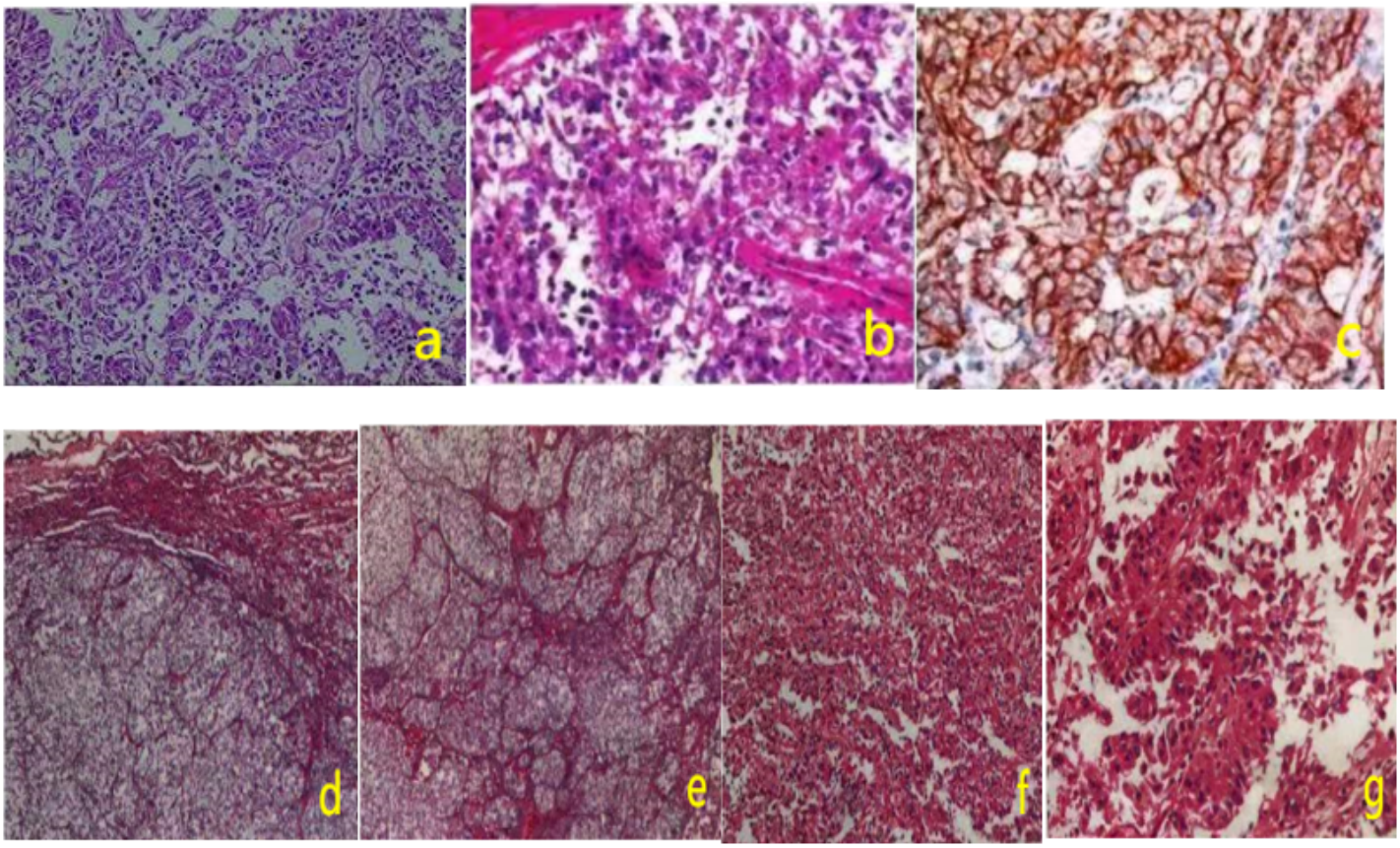

Figure 5

a: sigmoid colon and ovarian tissue: the tumor cells were arranged in solid nests, glandular tubes and papillae. The cytoplasm of tumor cells is transparent or eosinophilic; Figure b: immunohistochemistry of ovarian masses: CK pan (-), EMA (+), HMB45(+), Melan-A (-), E-cadherin (+), calretinin (-), CD117 (+), vimentin (-), NSE (+) SALLl4 (-), CD34 (vascular positive). Figure c: immunohistochemistry of ovarian masses: CK8/18 (-), CD99 (weakly positive), Inhibin-a (-), CK pan (-), β-catenin (+). Fig. d and e: immunohistochemistry of pulmonary nodules TFE-3 (+), HMB45 (+); Fig. f and g: immunohistochemistry of Tongji Hospital shows tumor cells: EMA (+), HMB45 (+), TFE-3 (+), E-cadherin (part positive), CyclinD1 (+), β- Catenin (some cell membranes were positive), Ki67 (Li: 1%); PCK, CK7, CK8/ 18, PAX8, SMA, Syn, Cg A, Cathepsin K, CD56, Mela A, S100, SOX10, Desmin, CD10, INSM1 and Vimentin (-).