

# Alveolar soft part sarcoma presenting as a uterine polyp: A case report

SAGE Open Medical Case Reports  
Volume 8: 1–4  
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DOI: 10.1177/2050313X20910598  
journals.sagepub.com/home/sco



Mariangela Gomez , Kerry Whiting and Rana Naous

## Abstract

Alveolar soft part sarcoma is a rare malignant soft tissue tumor, mainly localized in the extremities and occurring principally in adolescents and young adults. Alveolar soft part sarcoma are uncommon in the female genital tract. We here report a case of alveolar soft part sarcoma in a 20-year-old nullipara, presenting with vaginal bleeding and profound anemia requiring blood transfusions. Ultrasonographic examination revealed a polyp in the lower uterine segment. Surgical resection of the polyp was performed, and pathological evaluation showed typical histological, immunohistochemical, and molecular features consistent with alveolar soft part sarcoma. Patient underwent for total hysterectomy. Currently, she follows up with her surgeon and has no new complains. Given the infrequency of alveolar soft part sarcoma, this case report raises the awareness of alveolar soft part sarcoma as one of the entities to consider when confronted with a uterine polyp in a young patient.

## Keywords

Alveolar soft part sarcoma, uterine polyp

Date received: 3 July 2019; accepted: 30 January 2020

## Introduction

Alveolar soft part sarcoma (ASPS) is an indolent slowly growing malignant tumor that was first defined by Christopherson et al.<sup>1</sup> as a neoplasm of uncertain differentiation composed of polygonal cells arranged in an architectural nested pattern reproducing the alveoli of the lung, separated by fine fibrous septa and containing sinusoidal vascular channels lined by flattened endothelium. It usually occurs in adolescents and young adults and most commonly involves the extremities.<sup>2</sup> ASPS are rarely described in the female genital tract.<sup>3–6</sup> The prognosis of ASPS depends on the tumor stage. Patients with localized disease at presentation have a 71% 5-year survival rate, compared with 20% for patients with metastatic disease at time of diagnosis.<sup>7</sup>

Herein, we report a case of ASPS in a 20-year-old female, presenting with vaginal bleeding and profound anemia secondary to a lower uterine segment polyp that was later histologically proven to be ASPS.

## Case report

This is a case of a 20-year-old female who presented to her physician with complaints of continuous and heavy vaginal bleeding despite being on oral contraceptive pills. After a syncopal episode, the patient was found to be acutely anemic, requiring blood transfusion. On physical exam, she was

found to have a “small mass protruding from the cervix.” Pelvic Doppler Ultrasound (U/S) revealed a 3.5 cm x 3.5 cm x 2.5 cm oval echogenic mass with prominent vessels located within the cervix and extending from the fundus, compatible with a lower uterine segment polyp (Figure 1).

She further underwent D&C with polypectomy and specimen was sent for pathological examination. Grossly, the polyp had a tan-pink friable cut surface. Microscopic examination revealed an epithelioid neoplasm arranged in a nested pattern present within a highly vascular, edematous and fibrous stroma. The neoplastic cells displayed an abundant granular eosinophilic cytoplasm with prominent enlarged round nucleoli (Figure 2(a) and (b)). Immunohistochemistry (IHC) was performed showing strong nuclear positivity for TFE-3. The cells also expressed p53, INI-1, SMA (rare), and cathepsin-K (Figure 3(a) and (b)). Periodic acid–Schiff stain (PAS) highlighted intracellular crystalline material with the cytoplasm (Figure 3(c) and (d)). Fluorescence in-situ hybridization (FISH) studies showed evidence of TFE-3 and ASPSCR1 gene rearrangements. The tumor cells were

Department of Pathology, SUNY Upstate Medical University, Syracuse, NY, USA

### Corresponding Author:

Mariangela Gomez, Department of Pathology, SUNY Upstate Medical University, Syracuse, NY 13210, USA.  
Email: gomezma@upstate.edu

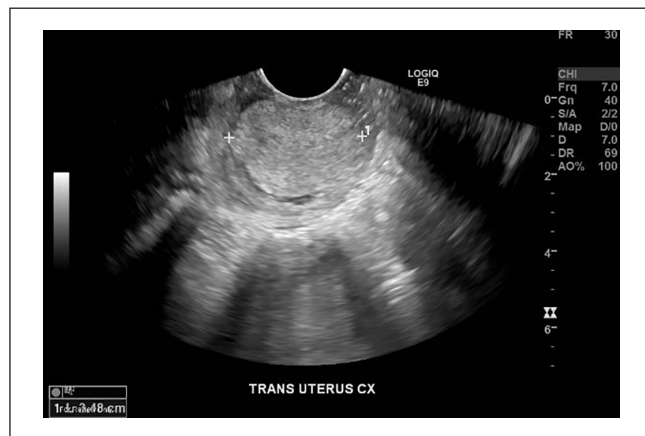


negative for epithelial markers, melanocytic markers, Oct 3/4 and SALL4. Given the overall morphology along with the immunophenotype and FISH results, the overall findings were consistent with the diagnosis of ASPS.

After exhaustive physical examination and imaging studies, clinical suspicious for metastatic disease by the surgical team was low. Patient underwent hysterectomy with lymph node sparing and her clinical status at the time of this publication has been uneventful. Currently, she follows up with her surgeon and has no new complains. No new masses or compromise of her health has been reported.

## Discussion

The first case of ASPS occurring in the female genital tract was reported in 1976 by Tobon et al.<sup>8</sup> which involved a 57-year-old woman with multiple vaginal nodules measuring

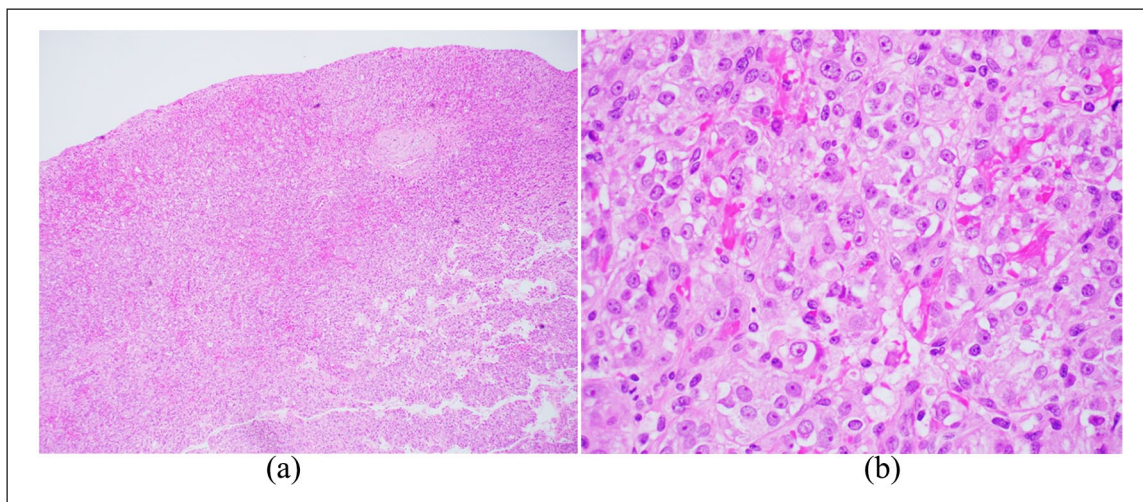


**Figure 1.** Pelvic Doppler U/S: echogenic mass within the lower uterine segment.

up to 0.8 cm. The largest series of ASPS involving the female genital tract was reported by Nielsen et al.<sup>9</sup> and involved nine patients with a mean age of 29 years presenting with abnormal vaginal bleeding and painless lesions measuring up to 9.8 cm. These tumors were most commonly located in cervix/lower uterine segment and uterine corpus (3 cases each), followed by the vagina (2 cases), and one case corresponding to the broad ligament.

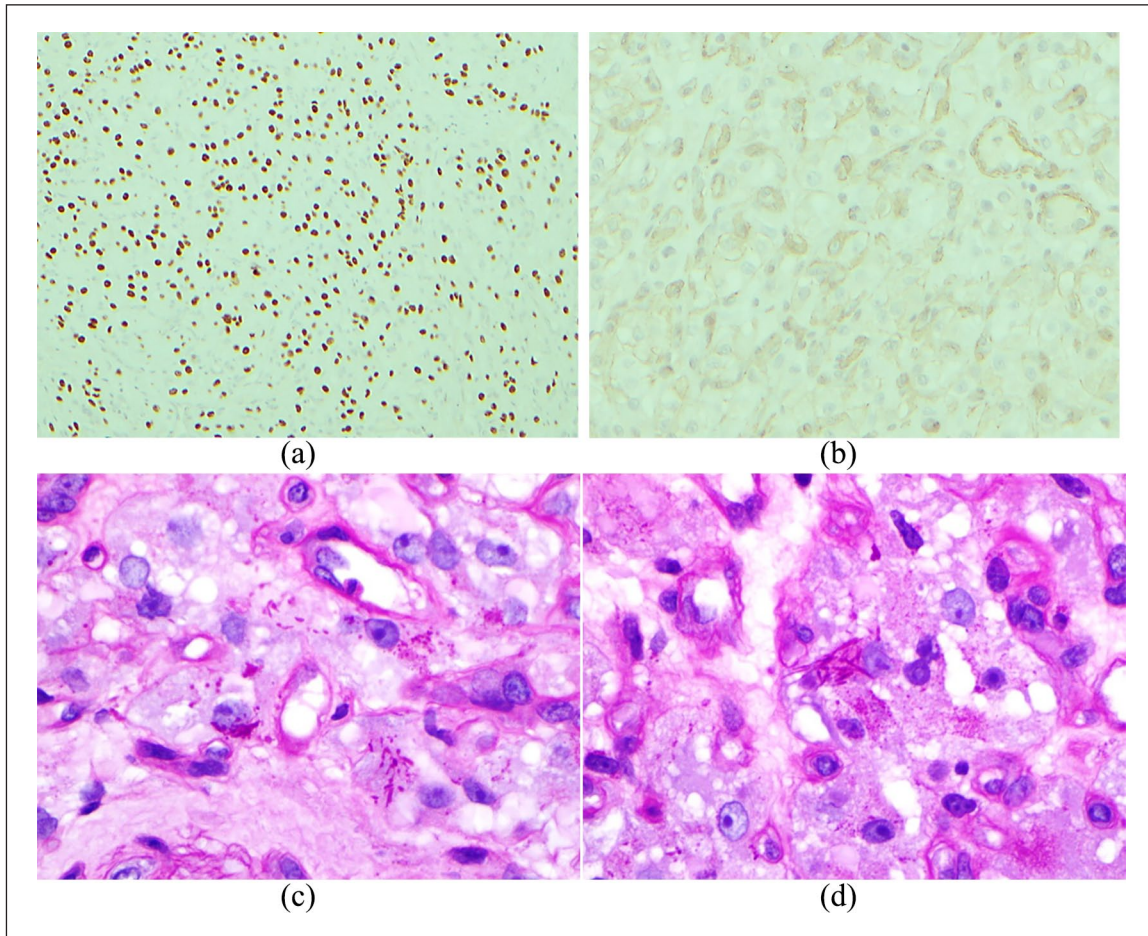
Grossly, ASPS of the genital tract presents as a poorly circumscribed tumor with a tan to yellow cut surface, and a soft to rubbery consistence. Foci of hemorrhagic degeneration could be seen, especially in large tumors. Tumor size is usually small and ranges from 2 mm to 4 cm, which might contribute to its relatively favorable prognosis compared with larger lesions in other locations.<sup>10</sup> Microscopically, ASPS presents as alveolar nests separated by delicate fibrovascular septae and characterized by a uniform population of medium to large polygonal cells with a prominent, single and centrally located nucleus, as well as eosinophilic granular cytoplasm which in most cases harbors characteristic crystalline inclusions that are usually visible with PAS-D stain or electron microscopy. Although mitotic figures are uncommon, vascular invasion is often present.

Strong nuclear TFE3 staining is considered a distinctive diagnostic marker for this entity. This phenomenon is a result of a recently defined and characteristic cytogenetic abnormality of der(17)t(X;17) (p11; q25) which fuses the TFE3 transcription factor gene on Xp11 to ASPL gene on 17q25. FISH and/or reverse transcription polymerase chain reaction (RT-PCR) are frequently used to confirm the presence of this ASPL-TFE3 fusion gene.<sup>11</sup> ASPS can also be positive for desmin, vimentin, cathepsin-K<sup>12</sup> and more uncommonly for S-100. It's consistently negative for other neuroendocrine markers such as synaptophysin and chromogranin A, epithelial markers such as cytokeratins and epithelial membrane antigen



**Figure 2.** (a) Hypercellular stroma with a nested tumor pattern separated by vascular channels, fibrous trabecula and edema (H&E, low power magnification). (b) Neoplastic cells arranged in an organoid/nested pattern showing a polygonal shape, eosinophilic, abundant and granular cytoplasm, with vesicular chromatin and prominent single nucleoli (H&E, medium power magnification).





**Figure 3.** (a) Neoplastic cells showing strong positivity for TFE-3 stain. (b) Weak staining for SMA is shown here. (c and d) Periodic acid–Schiff stain (PAS) highlighting the characteristic crystals (high power magnification).

(EMA), and melanocytic markers such as HMB45 and Melan-A. Although ASPS has an uncertain histogenesis, this immunoprofile is very helpful in distinguishing this tumor from other malignancies presenting with similar morphological characteristics.

Epithelioid smooth muscle tumors arising in the female genital tract usually present microscopically as nested to sheet-like proliferation of epithelioid cells with clear cytoplasm mimicking ASPS.<sup>13</sup> A transition to typical smooth muscle cells in most instances confirms the smooth muscle nature of these tumors along with their diffuse positivity for actin and desmin.<sup>14</sup>

Similar to epithelioid smooth muscle tumors, malignant rhabdoid tumor can mimic ASPS,<sup>15</sup> showing solid sheets of large cells with deep eosinophilic cytoplasm and eosinophilic hyaline cytoplasmic inclusions, but, unlike ASPS, has vimentin and cytokeratins positivity and absent INI-1 nuclear staining.

Perivascular epithelioid cell tumor (PEComa)<sup>16</sup> can display variable amounts of spindle, lipid laden or epithelioid cells with clear to eosinophilic cytoplasm with a centrally

placed and prominent nucleus. However, their positivity for HMB-45, Melan-A, and smooth muscle markers excludes ASPS.

Germ cells tumors, especially dysgerminoma, given the young age of the patient are a consideration in this case. However, the absent tumor staining for epithelial markers, Oct3/4 and SALL4 argued against such diagnosis.

Clear cell sarcoma<sup>2</sup> is also a very aggressive tumor with an indolent course presenting in young adults, and characterized by the presence of wreath-like multinucleated giant cells, intracytoplasmic melanin pigment (in 2/3 of the cases) and S-100, HMB45, and microphthalmic transcription factor (MITF) positive cells. On the other hand, ASPS is negative for all melanocytic markers.

Metastatic renal cell carcinoma, paraganglioma, and granular cell tumor serve as potential considerations in the differential diagnosis of ASPS; however, the young age of the patient, absent epithelial markers, neuroendocrine markers and S100 immunostain excluded these entities.

Finally, although extremely rare, metastatic ASPS to the uterus should be taken into consideration. Clinician should

exclude the presence of a primary lesion in the deep soft tissues of other parts of the body such as extremities, head, and among others. This has been reported by Lieberman et al.<sup>17</sup> and Portera et al.<sup>7</sup> as a critical factor to predict prognosis and survival rates.

## Conclusion

ASPS is uncommon in the female genital tract and could be mistaken for other entities. It is imperative to perform a complete histological examination and immunohistochemical workup as well as supporting cytogenetics studies in order to establish an accurate diagnosis.

## Declaration of conflicting interests

The author(s) declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

## Ethical approval

Our institution does not require ethical approval for reporting individual cases or case series.

## Funding

The author(s) received no financial support for the research, authorship, and/or publication of this article.

## Informed consent

Written informed consent was obtained from the patient(s) for their anonymized information to be published in this article.

## ORCID iD

Mariangela Gomez  <https://orcid.org/0000-0001-7346-201X>

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