

Epithelioid angiomyolipoma of the uterus: a case report and review of literature.

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Abstract

Background: Epithelioid angiomyolipoma (EAML)-a recently recognized pathologic entity-reportedly can develop at various anatomical sites, but rarely in the gynecological region, particularly in the uterus. **Case presentation:** We present a rare case of extrarenal EAML that developed within the uterus of a 57-year old woman without tuberous sclerosis. Magnetic resonance imaging (MRI) showed that the tumor was mainly composed of mature adipose tissue. **Conclusion:** This case offers new insight into the appearance of extrarenal EAML in the uterus on MRI.

Keywords: Epithelioid angiomyolipoma, Magnetic resonance imaging, Uterus.

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Introduction

Extrarenal epithelioid angiomyolipoma (EAML) has only been recently identified as a pathologic entity [1]. The occurrence of epithelioid AML in the female genital tract without tuberous sclerosis or tuberous sclerosis complex (TSC) is rare. In fact, only a few cases with this condition in the uterus have been reported this far, including a patient regraded as having hamartoma of the uterus by McKeithen et al. [1], another patient regarded as having lipoleiomyoma by Jacobs et al., and another patient considered as having lipomatous neometaplasia by Sieinski, in addition to 2 other cases [2,3]. Renal AML is known to be commonly associated with tuberous sclerosis (TS), wherein the tumor cells exhibit HMB45 immunoreactivity. The unique features of this neoplasm have been assessed by several pathologists in an attempt to define its pathogenesis. In the present report, we describe the clinicopathological and immunohistological features of a case with a uterine EAML and review the published literature on this topic.

Case Presentation

In July 2016, a 57 year old woman was admitted to our hospital for the evaluation of lower abdominal pain. Physical examination indicated a hard and well-demarcated mass. Routine laboratory tests yielded normal results. Subsequently, abdominal magnetic resonance imaging (MRI) was performed

using a GE 1.5 T LightSpeed scanner (GE Medical Systems, Milwaukee, WI, USA), which revealed a mildly lobulated, well-defined homogeneous fat-like mass, $8.4 \times 10.9 \times 14.1$ cm in size, in the left lower abdominal cavity. The mass exhibited high signal intensity on T1- and T2-weighted images, and low signal intensity on fat-suppressed T2-weighted images with multiple thin septa, which demonstrated slight enhancement on post-gadolinium administration (Figures 1A-1D).

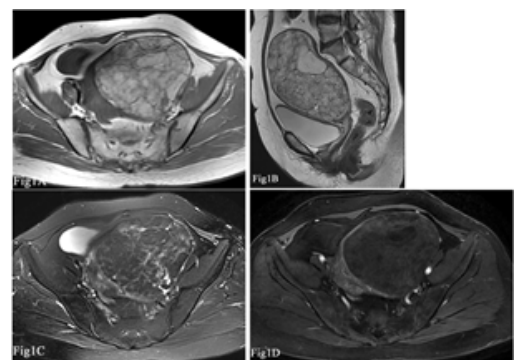


Figure 1. Magnetic resonance imaging (MRI) of the pelvis (a) Axial and (b) sagittal T1-weighted MRI shows a well-defined, slightly lobulated fat-like mass with high signal intensity in the uterus. (c) Axial fat-suppressed T2-weighted imaging reveals a mass with low signal intensity and multiple thin septa. (d) Axial contrast-enhanced T1-weighted MRI shows septa with light enhancement on post-gadolinium administration.

Brain imaging scans appeared normal. On laparotomy, circumscribed subserosal and intramural yellow lesions on the corpus uteri were noted and completely resected. On macroscopic examination, the mass was composed of an admixture of yellow mature adipose tissue, blood vessels, and smooth muscle, with an incomplete capsule and multiple fibrotic septa. In addition, immunohistochemical assays were performed using the following antibodies: Human Melanoma Black-45 (HMB-45), Melan-A, smooth muscle actin (SMA), S-100, cytokeratin, and Fish test. The tumor was positive for Melan-A (Figure 2) and SMA, and was negative for HMB-45, S-100, cytokeratin, and Fish test with MDM2 amplification. Based on these findings, the patient was finally diagnosed as having a uterine epithelioid AML.

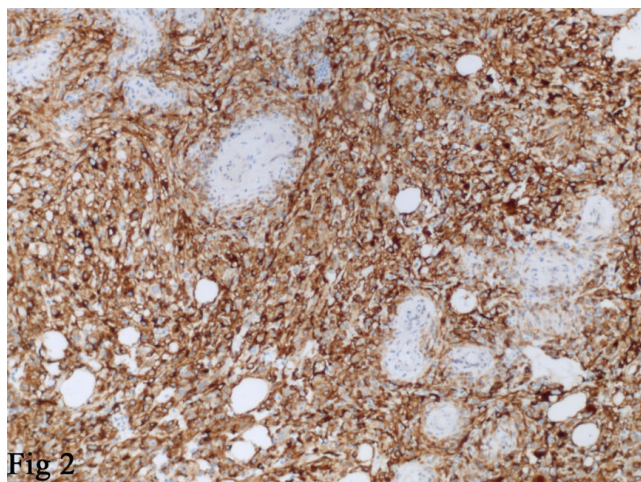


Figure 2. Melan-A immunostaining of the tumor. Strong and diffuse positive staining for Melan-A is noted in the tumor cells (100X).

Discussion

AML is a very common renal clonal mesenchymal tumor, and accounts for 2-6.4% of all renal tumors [3-5]. It had been regarded as a hamartoma rather than as a true neoplasm with malignant potential for a long time, because it is composed of an admixture of mature adipose tissue, thick-walled vessels, smooth muscle, and perivascular spindle cells of varying proportions within the tumor. Primary extrarenal AML is distinctly uncommon, and usually develops in the liver, nasal cavity, abdominal wall, and fallopian tube, according to previous reports [6-9]. The World Health Organization (WHO) defines this condition as a family of neoplasms termed as perivascular epithelioid cell tumors (PEComas), which can include renal or extrarenal AML, lymphangioliomyomatosis and clear cell tumors of diverse sites. AML may be categorized as sporadic and syndromic (accompanying TS or TSC) types [4,9]. The sporadic type of AML is the most common (80-90% of cases), and usually develops in older patients with a female predominance [6,7]. TSC is a group of autosomal dominant genetic disorders caused by germ-line mutations in the TSC1 and TSC2 genes, located on chromosomes 9q and 16p encoding the proteins hamartin and tuberin, respectively; this condition is characterized by the presence of hamartomatous lesions in multiple organs with no gender predilection [4].

AML can also be classified as classic (regular) and epithelioid, and the latter can also be further classified as epithelioid with atypia and epithelioid without atypia [10]. Classic AML is characterized by the presence of a variable proportion of mature fat tissue, smooth muscle cells, and thick-walled vessels, whereas epithelioid AML commonly exhibits the proliferation of predominantly epithelioid cells; in particular epithelioid with atypia may show aggressive behavior with malignant potential. Most of the cases of extrarenal AML are of the classic type and lack epithelioid components. In the present case, the tumor consisted of varying amounts of mature adipose tissue, hyalinized thick-walled blood vessels, smooth muscle components, and epithelioid components. Immunohistochemical characteristics, such as positivity for HMB-45, Melan-A, and SMA, can also help in the diagnosis of such uterine AML, although some extrarenal AML can also be negative for HMB-45 [6], as in the present case.

Renal or extrarenal AMLs can also be diagnosed *via* imaging, such as computed tomography or MRI alone. In fact, MRI is an important diagnostic tool for AMLs, as the proportion of fat within the lesion defines its suitability for imaging. Macroscopic fat has a negative CT value of <20 Hounsfield units (HU), which is a widely accepted cut-off for confirming the presence of fat and consequently the diagnosis of AML. Moreover, high signal intensity on T1 and T2 sequence imaging is associated with high sensitivity in the detection of extrarenal AML.

Extrarenal AMLs usually present with clinical features similar to those of a lipoma, lipoleiomyoma, angiolipoleiomyoma, and liposarcomas. The liposarcomas are most likely of choriomatous nature, and uterine lipoma, lipoleiomyoma, angiolipoleiomyoma probably arises from lipomatous metaplasia of a pre-existing leiomyoma, which are all included in the differential diagnosis.

The spectrum of clinical presentation in cases of uterine AML is broad, and depends on the tumor location and size. A previous study found that 70% of AML cases are symptomatic, whereas the remaining patients develop symptoms such as abdominal pain and mass effects [3,11]. Moreover, tumors occur with equal frequency in both sexes, although some studies show a male predominance.

In the present report, we describe a rare case of extrarenal AML in the uterus, and believe that AML should be included in the differential diagnosis of adipose layer lesions.

Conclusion

In summary, we present a case of AML without hemorrhagic, necrotic, or cystic components on MRI. We recommend that radiologists should be aware of the imaging characteristics of AML. This rare case offers new insight into the appearance of AML on MRI.

Acknowledgement

Written informed consent was obtained from the patient for publication of this case report.

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