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Imaging Manifestations and Pathological Basis of One Case of Alveolar Soft Tissue Sarcoma of the Gluteus Maximus Muscle

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Abstract: Alveolar soft tissue sarcoma is a rare malignant tumor of soft tissue, more common in young women, with deep soft tissues in the limbs and buttocks being the most prevalent sites. There are few reported cases in clinical practice. The clinical manifestations lack specificity and the imaging signs are diverse. This case presents ultrasound, MRI and PET/CT images of alveolar soft tissue sarcoma of the gluteus maximus muscle to enhance readers' understanding and awareness of the imaging signs of this rare disease in order to raise awareness of its diagnosis. The characteristics of this case are summarized and reported in combination with domestic literature.

Keywords: Alveolar soft tissue sarcoma; Gluteus maximus; Magnetic resonance; Positron emission tomography

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1. Introduction

Alveolar soft part sarcoma (ASPS) is a rare malignant soft tissue tumor characterized by an unknown tissue origin and tumor cells arranged in an alveolar or organoid pattern. The disease was first reported by Christopherson *et al.* [1] in 1952 and is prevalent in adolescent females, with deep muscles or fascia of the extremities being more common. Clinically, it often presents as a slow, painless mass, but due to its metastatic potential, it can spread to the lungs, bones, brain, subcutaneous tissue, etc. At present, radical surgery is the main treatment method, and regular follow-up monitoring of metastasis and recurrence is required after surgery. This article provides reference and assistance for clinical diagnosis and treatment by retrospectively analyzing the diagnosis and treatment process of one patient with gluteus maximus ASPS admitted to the Second People's Hospital of Guangdong Province in October 2024, combined with a literature discussion.

2. Clinical data

2.1. Basic information

The patient, male, 15 years old, was admitted to the hospital due to "a mass found in the right hip for more than half a year." Physical examination: A 4 cm × 5 cm mass was palpated in the right hip, soft in texture, without tenderness, with acceptable range of motion, and no local skin temperature increase. Over the past six months, the mass has grown slowly and progressively without tenderness, ulceration or nerve compression.

2.2. Imaging examination

2.2.1. Ultrasound examination

A hypoechoic mass, approximately 64 mm × 56 mm × 38 mm in size, with a clear boundary and shallow lobulation, and uneven internal echo was observed under the right hip. The mass was surrounded by tortuous and dilated veins. Color Doppler flow imaging showed abundant strip-shaped blood flow signals within the lesion, and the arterial spectrum was of low resistance type (**Figure 1** and **Figure 2**).

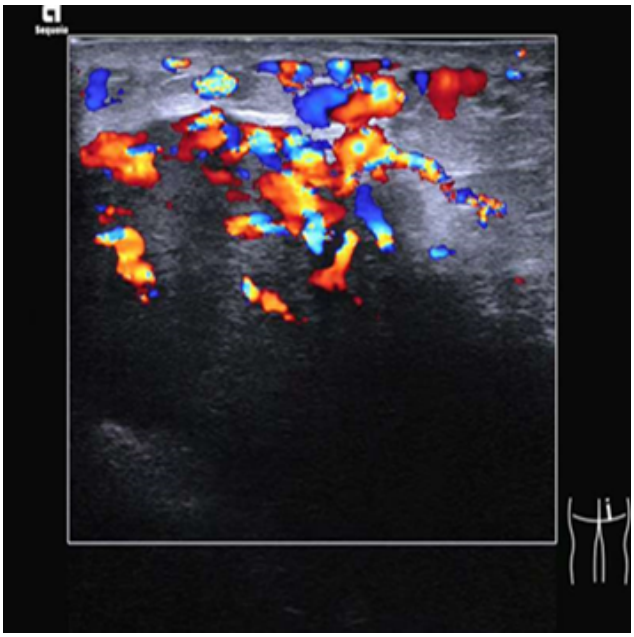

Figure 1. Ultrasonic image.

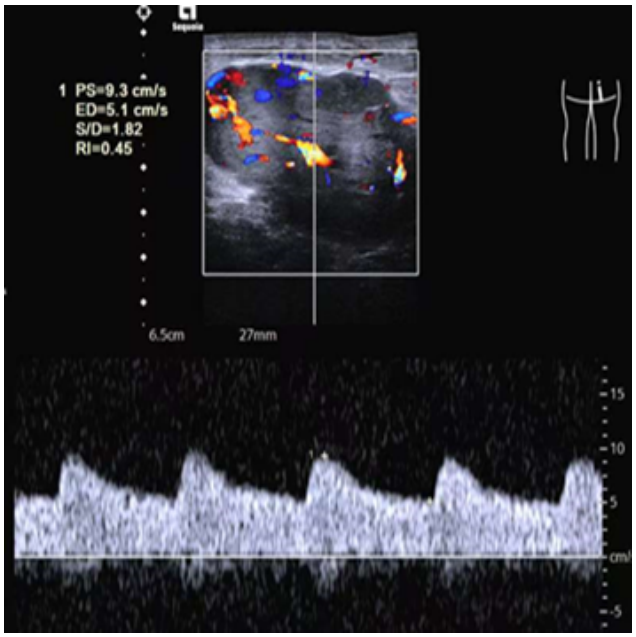

Figure 2. Ultrasonic blood flow spectrum.

2.2.2 Magnetic resonance imaging (MRI)

An abnormal signal space-occupying lesion was seen at the posterior margin of the right gluteus maximus muscle, with a clear boundary, closely related to the gluteus maximus muscle, approximately 38 mm × 71 mm × 68 mm in size. The T1WI sequence shows isosignal, with multiple patchy/cord-like low signals (**Figure 3**). The T2WI/fat inhibition sequence isosignaling, with multiple patchy hypersignaling within (**Figure 4** and **Figure 5**). A high signal in the isosignal area of DWI (Figure 6) and a low signal in ADC (Figure 7) suggest limited diffusion. The isosignal area was significantly enhanced after enhanced scanning, while the intrauterine strip area was not enhanced (**Figure 8**).

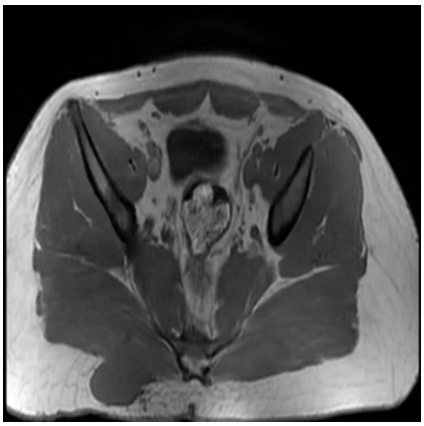

Figure 3. MRI T1WI image.

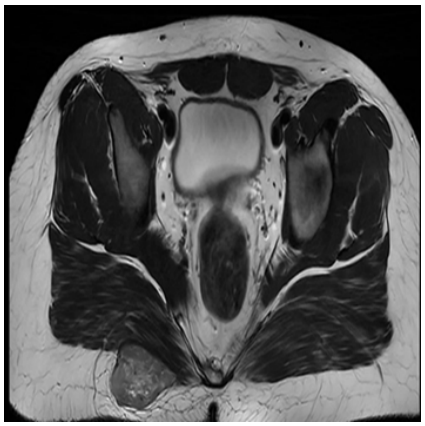

Figure 4. MRI T2WI image.

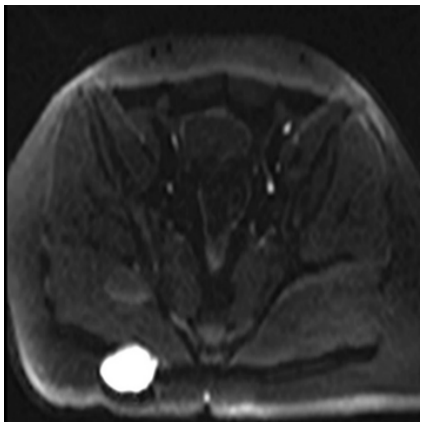

Figure 5. MRI T2-weighted imaging with fat suppression.

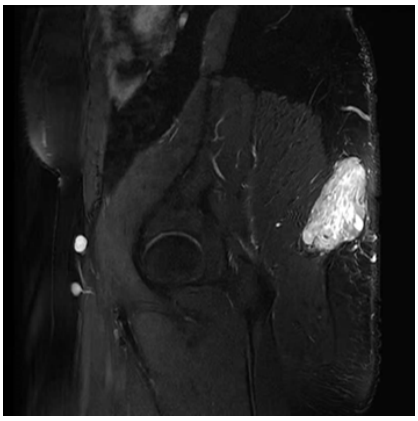

Figure 6. Diffusion weighted imaging, DWI.

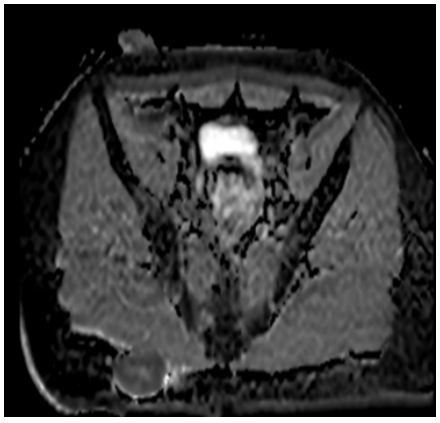

Figure 7. Apparent Diffusion Coefficient, ADC.

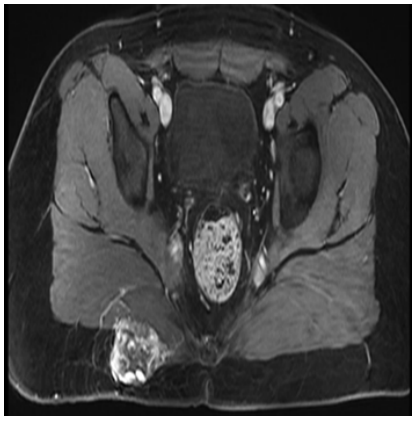

Figure 8. MRI enhanced scan image.

2.2.3. Positron emission computed tomography (PET/CT)

A soft tissue density mass was seen in the right buttock, with an indistinct boundary from the gluteus maximus on one side, approximately 50 mm × 27 mm × 65 mm in size, with abnormal FDG concentration, SUVmax of about 5.0. Multiple tortuous vascular shadows were seen subcutaneously in the lumbococcygeal region (**Figure 9** and **Figure 10**).

Based on the patient's age, medical history and imaging manifestations, mesenchymal malignant tumor was considered, with surgical indications, and surgery was proposed.

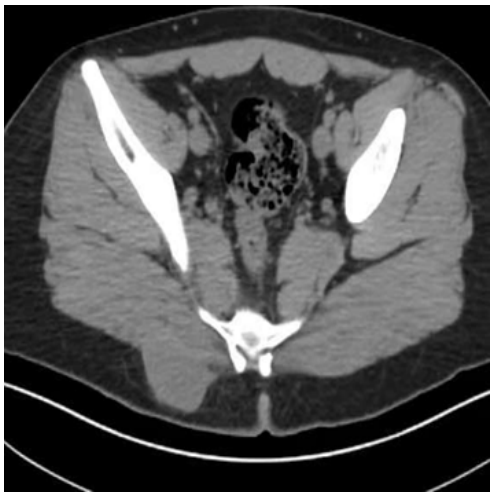

Figure 9. Plain CT scan image

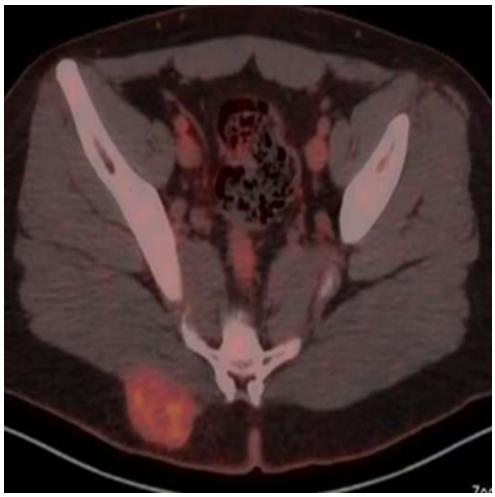

Figure 10. PET-CT image

2.3. Puncture and postoperative pathological results

The tumor tissue is arranged in an acinar pattern, with a rich network of slit-like capillaries in between. The cytoplasm of polygonal or large round tumor cells is rich in red staining, and some have nucleoli. Mitotic signs are rarely seen. Immunohistochemistry: CK/pan (-) EMA (-) Desmin (scattered few +) SMA (+) MyoD1 (-) Myogenin (-) TFE3 (+) S-100 (-) HMB45 (-) Melan-A (-) CD34 (interstitial vascular endothelium +) Ki-67 (about 3% to 5%+). Special staining: reticular fibers (showing acinar structures), D-PAS (showing a few PAS positive crystals in the cytoplasm), the lesion is considered to be alveolar soft part sarcoma (ASPS) (**Figure 11** and **Figure 12**).

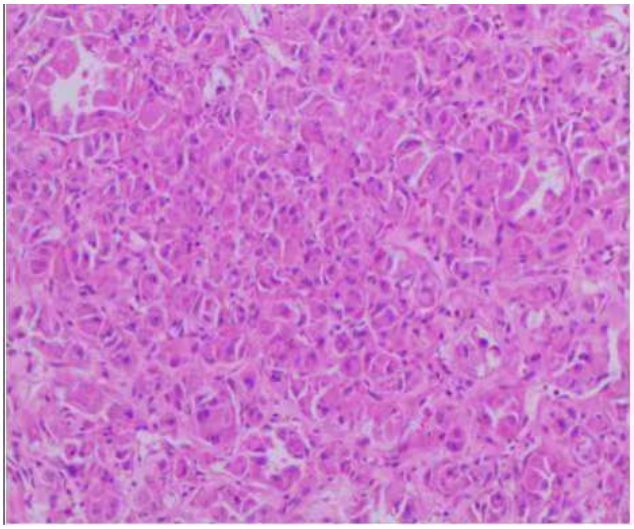

Figure 11. Puncture pathology.

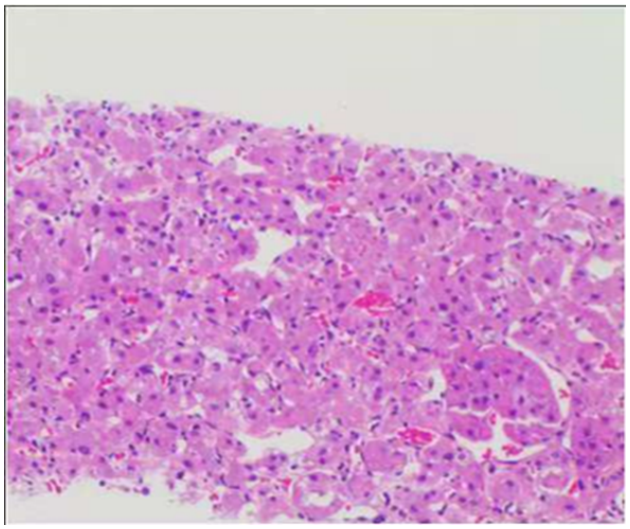

Figure 12. Postoperative pathological image.

3. Discussion

ASPS are malignant tumors with undetermined differentiation as defined in the 2020 WHO histological classification of soft tissue and bone tumors. Epidemiological studies have shown that the disease has a significant

gender bias and is more common in women aged 15–35 years old, especially in the deep muscles of the lower extremities and the buttocks. At the first diagnosis, the mass is often large, the tumor is deep, usually in the intermuscular space or between the muscle and the bone, and is not easily detected at an early stage [2]. ASPS grow slowly and often present as painless, progressively increasing masses, and are thus called "silent tumors." Although the histological grade is mostly FNCLCC 1-2, the rate of metastasis is relatively high, about 20 to 40 percent, most often to the lungs (about 42 to 65 percent), followed by the brain and bones [3]. The treatment strategy for ASPS is extensive resection, and postoperative chemotherapy may be crucial for long-term survival.

3.1. Imaging findings

On CT images, ASPS typically present as slightly low-density masses with relatively clear boundaries. CT scans can clearly show the location, size, and shape of the tumor, and whether there are features such as calcification, bleeding, or adjacent bone destruction. However, CT has limited value for qualitative diagnosis of tumors.

3.2. The diagnosis of ASPS relies more on MRI

MRI imaging features: On T1WI, ASPS often shows isosignal or slightly hypersignal, and their signal characteristics are closely related to the abundance of blood components within the tumor and the relatively slow blood flow [4]. In some cases, small patchy necrotic foci can be observed within the tumor, presenting as low-signal areas on T1WI and high-signal areas on T2WI [5]. On T2WI, the lesion often shows uneven high signal, reflecting the heterogeneity of vascular structure and the diversity of cellular components within the tumor, and the characteristic empty vascular shadow is significant within and around the tumor [6]. Dynamic contrast-enhanced scans showed significant heterogeneous enhancement of the lesion, and the enhancement mechanism involved the combined effect of dense vascular networks, arteriovenous fistulas, and larger "blood pool" volumes [7]. DWI showed a significant reduction in the apparent diffusion coefficient (ADC) value, suggesting the dual effect of high cell density and abundant buried tube structure in the tumor, which is positively correlated with the malignancy of the tumor [8].

3.3. Pathological features

ASPS are typically structured with characteristic alveolar or nest-like arrangements, and the cytoplasm is rich in PAS staining positive anti-amylase rhombic or rod-like crystals. Immunoexpression analysis showed strong positive expression of TFE3 in tumor cell nuclei, which has now become the gold standard for diagnosis [9]. At the molecular pathological level, ASPSCR1-TFE3 gene fusion is its specific molecular marker, and this genetic anomaly leads to the characteristic sinusiform vascular network structure in the tumor stroma by activating angiogenesis-related pathways (such as VEGF, MET). These vascular abnormalities are significantly associated with the therapeutic sensitivity of anti-angiogenic drugs, providing an important basis for molecular targeted therapy [10].

3.4. Differential diagnosis

ASPS need to be differentiated from the following soft tissue tumors:

(1) Muscle or intramuscular hemangioma: significantly high signal on T2WI (" bulb sign ") with clear boundaries; Common venous stones or calcified components are seen on CT, with significant nodular enhancement after enhancement, and the pathological basis is vascular endothelial cell proliferation and

thrombotic organization [11].

(2) Arteriovenous malformations: Dominated by vascular masses, with less solid components, and the image shows "earthworm-like" empty shadows formed by the thick supplying arteries and draining veins [12].

(3) Synovial sarcoma: It is more common in young adults (20–24 years old), and is often seen in the deep soft tissues beside the joint (such as the knee joint, foot); Imaging manifestations are mostly marginal calcification and necrotic cystic lesion areas, lack of ASPS characteristic empty vessels, and enhancement in patchy moderate enhancement.

(4) Myxomyfibrosarcoma: More common in the elderly (> 60 years old), mostly located beneath the superficial fascia; MRI shows "double low signal" (low signal on both T1WI and T2WI), and the pathology is mainly composed of collagen fiber bundles and mucinous matrix. After enhancement, the fiber components are significantly enhanced, and the mucinous area shows delayed enhancement [13].

4. Summary

ASPS is a rare malignant tumor with low incidence and diverse clinical manifestations, making accurate preoperative diagnosis quite challenging. Currently, the preoperative diagnosis of ASPS mainly relies on imaging examinations, but the imaging manifestations lack specificity and are easily confused with other soft tissue tumors, resulting in a high rate of misdiagnosis. Therefore, there is an urgent need to further summarize and refine the imaging features of ASPS by accumulating more cases to provide a more reliable basis for a clear preoperative diagnosis in the future.

Disclosure statement

The authors declare no conflict of interest.

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