Clinical Case Report

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Alveolar soft part sarcoma occurring in the penis of a 3-year-old boy

A rare case report

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Abstract

Rationale: Alveolar soft part sarcoma (ASPS) is a rare, malignant neoplasm, which mostly occurs in the upper and lower extremities. This article presents an unusual case of ASPS involving the penis of a 3-year-old boy. To our knowledge, this is the first case of ASPS in the penis of a child.

Patient concerns: The patient complained of slight penile pain for 1 year and a soft tissue mass could be palpated in his penis.

Diagnoses: Imaging was performed on the penis. The pathological feature of the mass was evaluated through biopsy examination. It was found that the mass was an alveolar soft tissue sarcoma, which was then confirmed by immunohistochemistry.

Interventions: The patient only underwent a partial penectomy because his parents wished to keep the penis. Conventional chemotherapy has been performed for 6 months after the surgery.

Outcomes: At 28-month follow-up the mass did not increase apparently, and no signs of metastasis were found.

Lessons: ASPS may occur originally in the penis.

Abbreviations: ASPL = alveolar soft part sarcoma locus, ASPS = alveolar soft part sarcoma, CT = computed tomography, MRI = magnetic resonance imaging, TFE3 = transcription factor E3.

Keywords: alveolar soft part sarcoma, pediatric, penis

1. Introduction

Alveolar soft part sarcoma (ASPS) is a type of sarcoma that constitutes approximately 0.5% to 1% of all soft tissue tumors.[1] It is a malignant, hypervascular neoplasm and shows a slight female predominant pattern.[2,3] Morphologically, its tumor cells are characterized by being arranged in an alveolar pattern; histochemical staining is useful for the diagnosis.[4,5] About one-third of ASPS occurs in children and adolescents.[6] It mostly occurs in the extremities, especially upper and lower extremities.[2] Other organs like the urinary bladder, breast, larynx, and the uterine cervix also have been reported.[7] The occurrence of distant metastasis is quite common, mainly to the lungs or brain.[8] Because the tumor usually runs a painless and slowly growing clinical course and rarely causes functional impairment, metastasis is the primary manifestation of the disease in some cases.[1,2] The 5-year overall survival rate can decline drastically in patients with metastasis.[2]

Here, we report a case of ASPS occurring primarily in the penis of a 3-year-old boy and present the clinical presentation, imaging findings, and treatment. To our knowledge, this is the first case of ASPS in the penis of a child.

2. Methods and results

Written informed consent was obtained from the parents of this patient for the publication of this report and accompanying photographs under the guidelines of the Medical Ethics Committee of the Huazhong University Graduate School and Faculty.

A 3-year-old boy who complained of slight penile pain for 1 year was admitted to our hospital on February 17, 2014. According to his parents, he had no urinary symptoms. Physical examination of this child revealed a swelling at the root of the penis, which was hard, immobile, and had some tenderness on palpation. Palpable inguinal lymph nodes were present, and the largest one was measured approximately 1 cm in diameter. No other positive findings were found, and there was no family or genetic history. The routine laboratory tests were within normal limits. Grayscale and color Doppler ultrasound demonstrated a well-circumscribed hypoechoic mass in the penis with hypervascularity (Fig. 1A and B). A subsequent magnetic resonance

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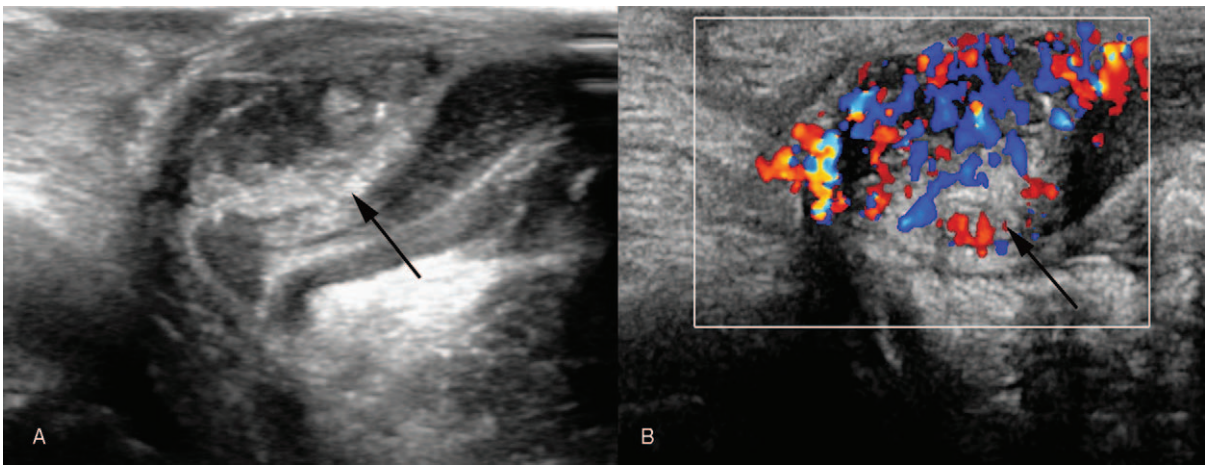

Figure 1. Ultrasonic findings. Grayscale ultrasound showed a well-circumscribed hypoechoic mass in the penis (A, arrow). Color Doppler image showed increased vascularity in the mass (B, arrow).

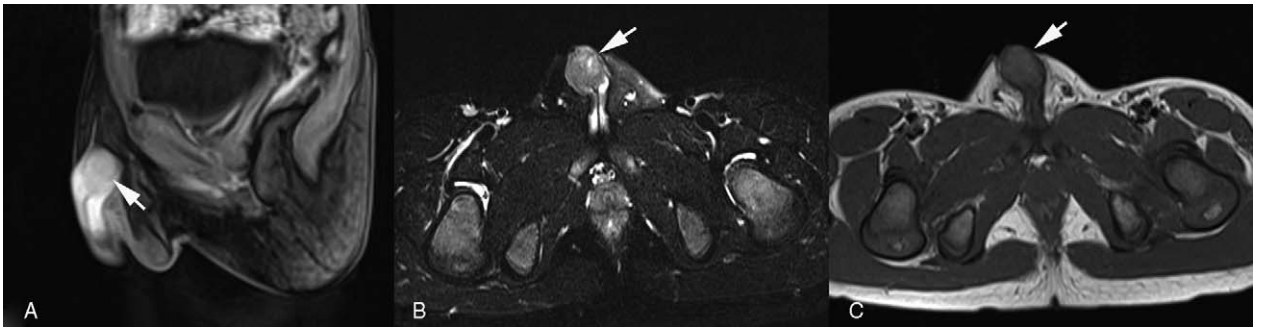

Figure 2. Contrast-enhanced magnetic resonance (MR) images of the penis demonstrated a well-enhanced mass with some large vessels involving the dorsal part of corpora cavernosa (A). Magnetic resonance imaging showed the mass appearing with slightly hyperintense on axial T2-weighted images (B) and slightly hyperintense on axial T1-weighted images (C).

imaging (MRI) demonstrated a mass showing slightly hyperintense on T1 and T2 in the dorsal part of the corpora cavernosa away from membranous urethra (Fig. 2B and C). The mass was measured $1.8 \times 1.6 \times 2.1$ cm. There was an intense enhancement and some large vessels in the region of the lesion when the mass was enhanced (Fig. 2A). The mass was also well enhanced on contrast-enhanced computed tomography (CT) scan and considered as hypervascular neoplasm.

This patient was planned to undergo an organ-sparing penectomy under general anesthesia. No signs of metastasis were found before an operation. However, after dissection, exploration showed the mass in the dorsal aspect of the penis occupied 4/5 volume of the corpora cavernosa and had an unclear border with the urethra and the surrounding tissue. Thus, only 2 pieces of tissue were taken from the tumor for histopathologic examination. The gross appearance of the mass was firm, and the cut surface was grey white. Pathological photomicrograph showed large, round to polygonal tumor cells containing eosinophilic granular and vacuolated cytoplasm (Fig. 3). A diagnosis of alveolar soft part sarcoma was rendered. Immunohistochemistry then confirmed it because of positive transcription factor E3 (TFE3) (Fig. 4). The postoperative course of this boy was uneventful, and he was discharged in good condition 10 days after the operation.

Considering that ASPS is aggressive and has the possibility of dissemination, further radical resection was suggested, but this may lead to penectomy. Given the young age of this patient, his

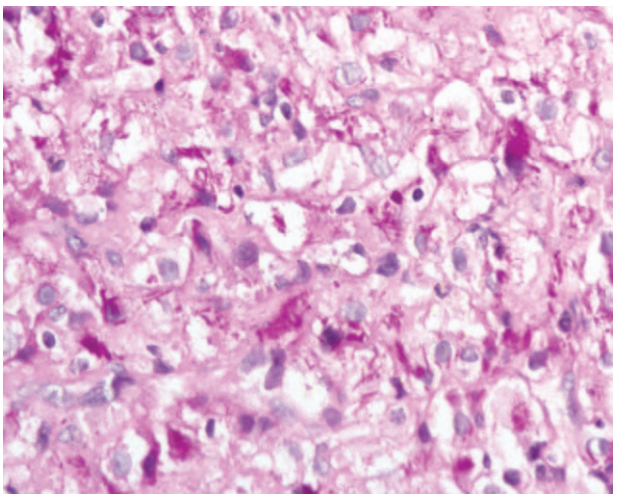

Figure 3. Photomicrograph (hematoxylin and eosin staining, ×400) showed large, round to polygonal tumor cells containing eosinophilic granular and vacuolated cytoplasm.

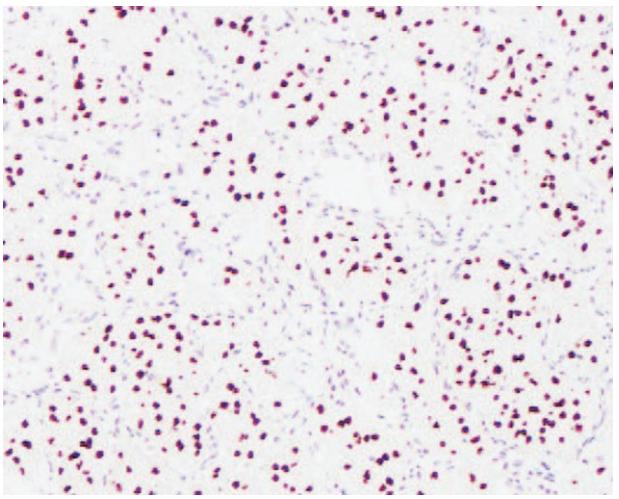

Figure 4. Positive immunoreactivity of TFE3 was observed (immunohistochemical staining, ×100). TFE3 = transcription factor E3.

parents rejected surgical intervention and opted for conservative medical therapy. A chemotherapy regimen was then started 2 months after surgery to debulk the tumor. The patient received 6 months of chemotherapy with ifosfamide, and epirubicin on 5 consecutive days of each month, mesna once at the first month, dacarbazine once at the third and fourth month. He tolerated well without any episode of febrile neutropenia except for emesis, which happened a few times during the chemotherapy. The size of the mass did not increase significantly after completion of the chemotherapy, and his parents refused to receive an additional cycle. No evidence of metastasis was found on his routine examination and whole-body bone scintigraphy at 28-month follow-up (Fig. 5).

3. Discussion

ASPS is a rare, malignant tumor and predominantly develops in young patients (<30 years old).[1,3] It often occurs in the deep soft tissues of the extremities, followed by the torso and the head and neck regions.[6] In this article, we reported a 3-year-old boy diagnosed with ASPS, whose primary tumor occurred in the penis. To our knowledge, no ASPS found in such position of a young patient has been documented in the literature. Furthermore, ASPS usually presents with a relatively indolent clinical course and causes no pain which made patients easy to neglect it.[1,2] However, in our case, the patient first complained of local pain, showing an unusual clinical presentation.

Histologically, ASPS is characterized by neoplastic cells containing eosinophilic granular or vacuolated cytoplasm, showing in an organoid pattern and most are associated with abundant sinusoidal vessels.[5] Molecular cytogenetic analysis can help the diagnosis of ASPS, because an ASPL-TFE3 (novel gene-transcription factor) translocation, which caused by a unique chromosomal rearrangement der(17)t(X;17)(p11;q25), has been identified.[5,9] As demonstrated in this case, immunohistochemical staining can identify the TFE3 target and confirm the diagnosis of ASPS.[4]

Imaging findings have an important accessory diagnostic value for ASPS. Ultrasonography is the initial imaging modality in our case. The ultrasonographic features are consistent with sonographic findings of ASPS occurring in other sites that had been reported before, which are well-defined hypoechoic mass and extreme hypervascularity.[10] Otherwise, the tumor exhibited large vessels and demonstrated intense contrast enhancement on CT and MRI.[11] These imaging findings are in agreement with the extremely vascular nature of ASPS, which is well correlated with the hypervascularity found in pathological specimens.[3,12] In contrast to others who have reported flow void of ASPS on MRI, we found no evidence of flow void in our case.[3] This

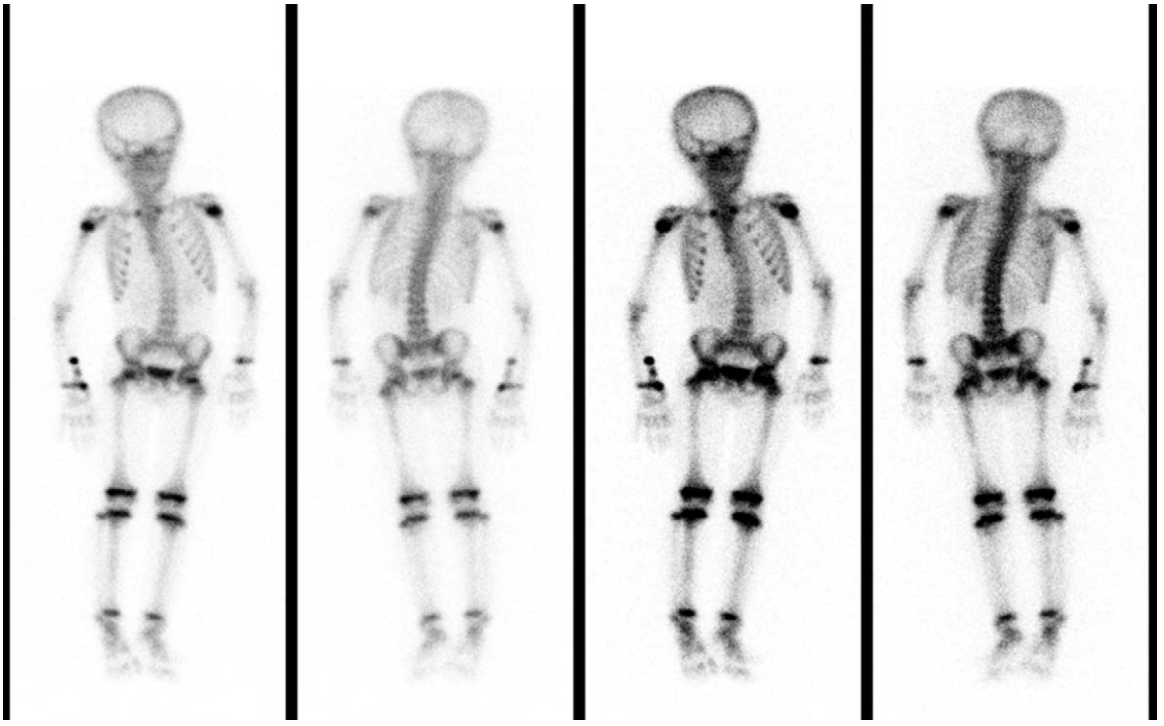

Figure 5. Bone scintigraphy did not detect increased or decreased uptake.

difference may due to our small sample size. Similar to prior reports, the tumor in our case exhibited slightly hyperintense on MR T1WI images and MR T2WI images.[3] This feature is in contrast to most pediatric soft tissue sarcomas which are hypointense on T1W images.[13]

Radical resection is associated with better prognosis in ASPS patients without metastasis. Due to the chemo insensitivity of ASPS, radical resection of the primary tumor and supplemental radiation treatment were suggested.[9,14,15] However, things became complicated in our case when the tumor occurred in the penis of such a young age patient. In consideration of the enormous social effect on his future life, radical resection and radiotherapy were refused by his parents. Fortunately, the follow-up examination showed no signs of new metastatic nodules and the patient did not complain penile pain anymore after therapy; 28 months of progression-free survival have been achieved. The young age and tumor size of our patient may contribute to his survival. Our patient was only 3 years old and the maximum diameter of the tumor was 2.1 cm. Previous studies suggested that children with ASPS have better 5-year survival than older patients and small tumor (size <5 cm) is associated with longer progression-free survival.[16] Reports regarding prognostic factors in ASPS also demonstrated that older age, large tumor (size >10 cm) are significant negative predictors of survival.[2]

However, ASPS has a propensity to metastasize, and distant metastasis is 1 predictor of poor outcomes.[1,6] In patients who present with the locoregional disease only, the 5-year overall survival rates were 82%, while in patients with metastasis, this decreases to 27%.[2] Some recent reports demonstrated that use of angiogenesis inhibitor to treat metastatic ASPS produced a satisfactory clinical outcome.[17,18] A histological analysis of ASPS cases suggested that anti-angiogenic therapy may help to prevent the metastasis, because it almost exclusively follows the invasion-independent pathway for entry into circulation.[19] Despite studies with a larger patient cohort are still required to validate the benefit, anti-angiogenic therapy may provide an additional conservative therapeutic option for ASPS, especially for patients with metastatic ASPS or those who need effectively conservative treatment just like the one in our case.

4. Conclusion

This case suggests that ASPS could be considered as a possible diagnosis, although it is rare occurring in the penis. Imaging feature is that ASPS present a hypervascular mass on ultrasonography, CT, and MRI. Pathology and immunochemistry are required to make a confirmative diagnosis. Radical resection is recommended to treat ASPS for good prognosis. However, we report a child of ASPS occurring primarily in the penis who had successful conservative approach.

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