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**Pathology**

# Rare Case of Alveolar Soft Part Sarcoma Presenting as a Malignant Bone Tumor: Case Report

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**Abstract** **Case Report**

Alveolar soft part sarcoma is a slowly growing malignant tumor with high potential of metastasis. It occurs most frequently in children and young adults. Cytogenetically, it is characterized by a specific translocation, der(17)t(X;17)(p11 ; q25). Because of its rarity, treatment is not yet standardized. The combination of surgery and radiotherapy remains the mainstay of the treatment.

**Keywords:** alveolar soft – part sarcoma, bone, metastases, TFE3.



## INTRODUCTION

Alveolar soft part sarcoma (ASPS) is a rare sarcoma accounting for less than 1% of all soft tissue sarcomas. It represents a high potential lesion that most commonly metastasizes to the lung, bone, and brain. An immunohistochemical study using TFE3 is useful in confirming the diagnosis and testifies the specific translocations (X;17). Treatment is not yet standardized because of its rarity. We present a case of a patient diagnosed with ASPS histologically but whose radiological image points rather to a malignant bone tumor.

## CASE REPORT

We report the case of 42 years old woman without any medico-surgical or trauma story. She consulted our structure for a painful knee. Physical examination found a mass in the upper extremity of the right leg. It was fixed, firm, and measures approximately 3 cm. the knee examination was unremarkable.

Magnetic resonance imaging showed a poorly-circumscribed mass of the proximal tibial epiphysis and metaphysis. This lesion leads to a rupture of the cortical and infiltrates the popliteus muscle. It has a high signal intensity on T2 (figure 1).

In front of this radiological aspect, a malignant bone tumor was suspected. The patient underwent a biopsy. Histopathologic examination revealed that the tumor is composed of large cells with abundant, eosinophilic cytoplasm and a round nucleus with a prominent nucleolus. Tumor cells were arranged in a uniform nest separated by fibrous septa containing sinusoidal vascular channels.

Immunohistochemical stainings showed that the tumor cells were positive for TFE3 but negative for cytokeratin, desmin, myogenin, chromogranin and synaptophysin and MelanA (figure 2). These pathology results were consistent with the diagnosis of an alveolar soft part sarcoma infiltrating bone. The patient subsequently benefited from wide resection of the tumor (figure3). A histological study of the resected specimen confirmed the diagnosis.

The patient was subsequently referred to the radiotherapy department.


***Figure1:*** Sagittal (A) coronal (B) and axial (C) views of contrast-enhanced magnetic resonance imaging showing a heterogeneously enhanced mass of the proximal tibial epiphysis and metaphysis. This lesion leads to a rupture of the cortical and infiltrates the popliteus muscle

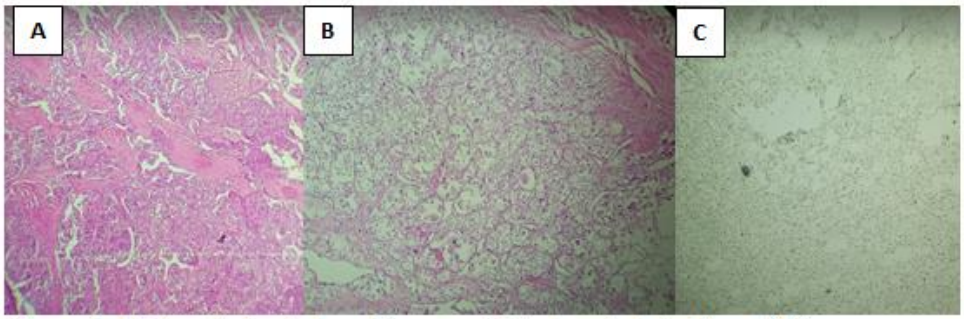


***Figure 2:*** **Histopathological and immunohistochemical studies: A)** Low-power view of **an** alveolar soft part sarcoma showing the pseudoalveolar growth pattern (a haematoxylin and eosin stain ×40).B) cells show abundant eosinophilic cytoplasm and vesicular nuclei with prominent nucleoli (a haematoxylin and eosin stain ×100). C) Strong an diffuse nuclear staining of TFE3 (_40).

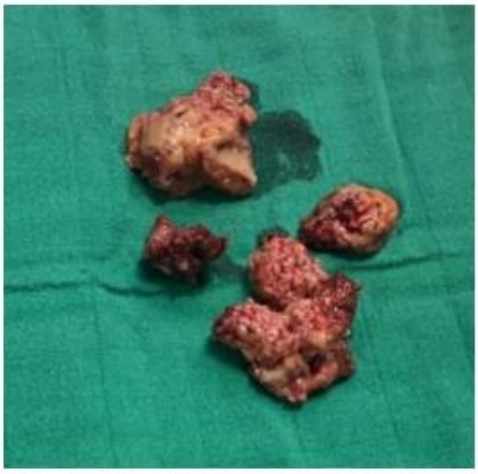

***Figure 3:*** macroscopic image of the resected mass

## DISCUSSION

Alveolar soft part sarcoma (ASPS) is a rare malignant tumor accounting for less than 1% of all soft

tissue sarcomas.1 [1]. It was first described as a distinct entity by Christopherson *et al*., in 1952.

This tumor occurs mainly in adolescents and young adults aged between 15 and 35 years. Young children can also be affected, but very few cases have been reported.2 [2]. There is a slight female predilection, especially in patients under 30 years [3]. In adults, ASPS arises most frequently in the lower extremities and trunk. In children, the head and neck, particularly the orbit and tongue, are the most common sites. Other more rare locations have been reported, namely the uttering cervix, penis and lung [4]. Patients with ASPS present generally with a non-ulcerated, painless mass that has gradually increased in size [5].

Other patients were consulted for clinical signs related to their metastases, such as Headache, and nausea in the case of brain metastases [4].

The radiological investigations, including ultrasonography, computed tomography (CT), and magnetic resonance imaging MRI reflect the hypervascular character of alveolar sarcoma [6]. Ultrasonography shows hypoechoic or a hyperechoic mass with extremely increased blood flow on CT images, the tumor appears as a mass with rich vascularity and an intense enhancement. On MRI, ASPS has characteristic findings, it presents high signal intensity on both T2- and T1-weighted images. Moreover, many blood vessels are observed in and around the tumor [5-7].

Gross examination of alveolar soft part sarcoma reveals a poorly circumscribed mass with friable consistency, tan-pale to yellow cut surface, and areas of necrosis and hemorrhage [8].

Histologically, the alveolar forme of ASPS consists of lobules with central loss of cellular cohesion, divided by thick fibrous septa containing a rich vascular network. Tumor cells are large, round to polygonal with well-defined borders, abundant eosinophilic granular cytoplasm, and a round nucleus with a prominent nucleolus. The Solid type of ASPS, is composed of clusters of cells without a nestlike pattern.

Histochemical stains using PAS are useful for diagnosis, it reveals intracellular glycogen and highlight characteristically PAS-positive diastase-resistant rod-shaped crystals. Immunohistochimacally, tumor cells show typically positive nuclear staining for TFE3 and cathepsin K. Desmin can be focally positive.

- The differential diagnosis of ASPS may occur with neoplasm showing, histologically, nested or organoid or cells with abundant eosinophilic cytoplasm. Malignant melanoma, renal cell carcinomas, paragangliomas and hepatocellular carcinomas are the main differential diagnoses. Immunohistochemical studies help in this distinction [9 – 10].
- Investigations of genetic abnormalities revealed that ASPS is associated with a specific translocation der(17)t(X;17)(p11;q25). It involves 2 genes, the TFE3 gene located on chromosome X (Xp11.2) and the ASPL gene located on chromosome 17 (17q25). The fusion gene that results from this translocation encodes for a fusion protein located in the nucleus and acts as an aberrant transcription factor [3]

The initiating genetic event in ASPS is ASPSCFI1-TFE3 translocation. The etiology of this translocation is still unclear. However, several risk factors including irradiation, genetic factors and chemical carcinogens have been described [1].

The optimal treatment for alveolar sarcoma is not yet standardized given their rarity. Complete surgical resection of primary soft tissue tumor and secondary location has been reported as the treatment of choice. Adjuvant chemotherapy does not seem to be effective because of chemo insensitivity of these tumors (11). The literature review reveals that radiotherapy may reduce the risk of local recurrence. It may be indicated in case of inadequate surgical resection or insufficient surgical margins. Recent studies have reported a significant benefit of tyrosine kinase inhibitors for ASPS [12].

ASPS have a high potential for metastasis. Lung, bone, and brain are the most common metastatic sites. Metastases to the lymph nodes remain rare. It can be diagnosed after more than 10 years [3].

ASPS is regarded as high-grade by definition [3]. Young age, tumor size of less than 5 cm and localized disease at the time of diagnosis are factors of good prognosis. Children with a tumor that arises in the head and neck have an excellent prognosis with 100 % survival rate of 5-years [8].

## CONCLUSION

Alveolar soft part sarcoma is a rare soft tissue sarcoma that occurs frequently in young patients. Early diagnosis and treatment with surgery alone or associated with radiation improves the prognosis and reduces the risk of recurrence.

**Cover letter**

Dear editor, we report a very interesting case of an alveolar soft part sarcoma which presented radiologically as an infiltrating malignant bone tumor.
I guarantee you, me ihssan elouarith (the author of this article) that:

- The current “Guide for Authors” has been read and our case report is compliance with the instructions and the conditions posed.
- All authors have seen and agreed to the submitted version of the paper, and bear responsibility for it.

- All who have been acknowledged as contributors or as providers of personal communications have agreed to their inclusion;
- The material is original and was not published elsewhere nor submitted for publication simultaneously.

If accepted, the paper will not be published elsewhere in the same or similar form, in English or in any other language, without written consent of the copyright holder.

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**Guarantor:** Elouarith Ihssan

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