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Pathology

ALVEOLAR SOFT PART SARCOMA OF THIGH - A CASE REPORT

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ABSTRACT Alveolar soft part sarcoma (ASPS) is a rare, slow growing soft tissue tumour of an undetermined histogenesis. It is among the least common sarcomas, representing 0.2-1% of large studies of soft tissue sarcomas. We report a case of a 35 years old female, who presented with a swelling in the medial aspect of her right thigh of 3 years duration, rapidly increasing in size over the past 3 months. USG guided trucut biopsy as well as Excision and HPE both were performed. Microscopy of which revealed round to oval tumour cells with abundant eosinophilic and granular cytoplasm , round to oval nuclei with vesicular chromatin and prominent nucleoli arranged in alveolar and nested pattern suggestive of Alveolar soft part sarcoma and confirmed by immunohistochemistry.

KEYWORDS : alveolar, sarcoma

INTRODUCTION

Alveolar soft part sarcoma is a clinically and morphologically distinct soft tissue sarcoma first defined and named by Christopherson et al.[1]in 1952. Since 1952, numerous examples have been reported and studied immunohistochemically and electron microscopically, but there is still uncertainity as to its exact nature and line of differentiation.They are uniformly malignant; there is no benign counterpart of this tumour. Alveolar soft part sarcoma is an uncommon neoplasm, estimated at 0.2%-1% of all soft tissue sarcomas.

CASE SUMMARY

A 35 year old woman presented with a slow growing mass in the medial aspect of her right thigh for three years. She had noticed a somewhat rapid increase in size since three months. Local examination revealed a large 4.8×4.5 cm painless mass in the posteromedial aspect of her right thigh. MR imaging showed presence of an intramuscular soft tissue sarcoma with multiple hypointense internal fibrous septae and prominent vessels [Figure 1]. Fine needle aspiration was done from the thigh mass which showed moderately cellular smears composed of large round cells with an abundant finely granular or vacuolated cytoplasm, markedly enlarged round nuclei with a prominent central nucleoli, and few scattered bare nuclei [Figure 2]. An impression of malignant small round cell tumour was made. USG guided trucut biopsy followed by excision and histopathological examination both were done.

PATHOLOGIC DETAILS:

On Gross examination, we received a solid, gray white, fleshy tumour mass measuring 4.5×4.3×3 cm , the cut surface of which was variegated in appearance showing whitish areas with scattered areas of necrosis and haemorrhage [Figure 3]. Microscopic examination showed tumour cells arranged in alveolar and nested pattern [Figure 4]. The tumour cells are round to oval with abundant eosinophilic and granular cytoplasm, round to oval nucleus with vesicular chromatin and prominent nucleoli [Figure 5].The closest muscle cut margin was free from tumour. Diagnosis of Alveolar soft part sarcoma was made; and the same was confirmed by immunohistochemistry, as the tumour cells showed nuclear positivity for transcription factor E3(TFE3) [Figure 6].

RADIOGRAPHIC FINDINGS

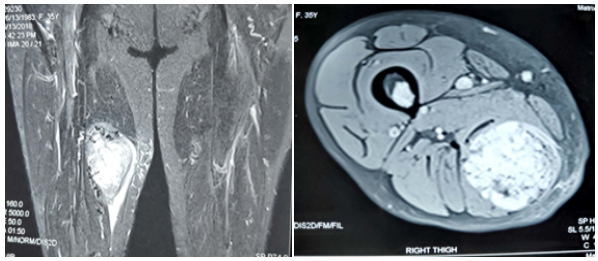

Figure 1: MR findings suggestive of intramuscular soft tissue sarcoma with multiple hypointense internal fibrous septae & prominent vessels

CYTOPATHOLOGICAL EXAMINATION

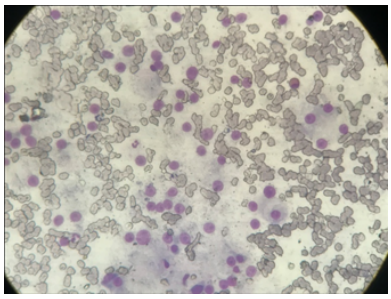

Figure 2: : cells having abundant granular cytoplasm and large round nuclei with prominent central nucleoli. Numerous stripped nuclei also seen.(MGG,400X)

GROSS EXAMINATION

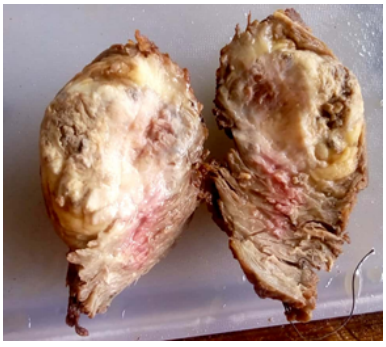

Figure 3: Cut surface of the tumour mass from the Right thigh showing fleshy, greyish-white, variegated appearance, along with some areas of necrosis along with attached muscle.

HISTOPATHOLOGICAL EXAMINATION

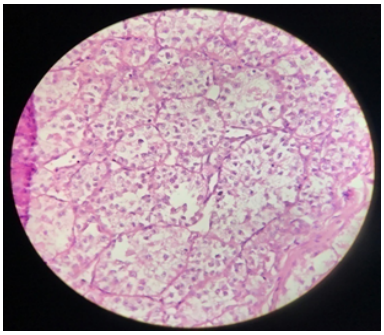

Figure 4: tumour cells arranged in alveolar and nested pattern; the nests being surrounded by thin walled vessels.(H & E,40X)

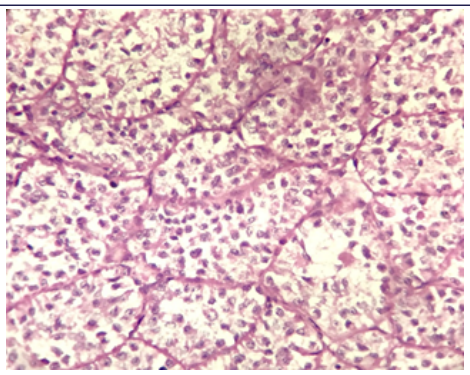

Figure 5: the tumour cells are dyscohesive,round to oval with abundant eosinophilic and granular cytoplasm.The nucleus is round to oval with vesicular chromatin and prominent nucleoli.(H&E,400X)

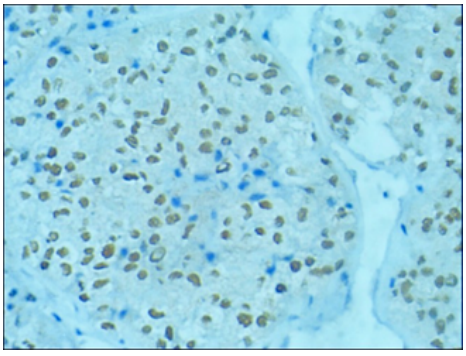

Figure 6: strong diffuse positive nuclear expression of Transcription Factor E3(TFE3) by the tumour cells.

DISCUSSION

Alveolar soft part sarcoma (ASPS) is a rare, distinctive sarcoma, typically occurring in young patients. Despite a relatively indolent clinical course, the prognosis is poor and is often characterized by late metastases and an extended clinical course; with survival rates of 77% at 2 years, 60% at 5 years, 38% at 10 years and only 15% at 20 years have been reported by Lieberman *et al*[2]. Since its formal description by Christopherson *et al* at Memorial Sloan Kettering Cancer Center, New York, USA[3]

ASPS has been the subject of considerable interest for pathologists and clinicians owing to its unique microscopic features, uncertain line of differentiation and unpredictable clinical behavior.

The tumour occurs principally in adolescents and young adults and is most frequently encountered in patients 15-35 years of age[4-6]. Female patients outnumber males, especially among patients under 25 years of age[7]Infants and children are affected less frequently. When it occurs in adults, it is seen predominantly in the lower extremities,especially the anterior portion of thigh. In a study of 102 alveolar soft part sarcomas by Lieberman et al.[4] , 39.5% involved the soft tissues of the thigh or buttock. The tumour has also been described in a variety of unusual locations, including the female genital tract[8], breast[9], urinary bladder[10] and bone[11]. In infants and children, orbit and the tongue are usually affected.

It usually presents as a slowly growing, painless mass that almost never causes functional impairment. Because of the relative lack of symptoms, it is easily overlooked; in a number of cases, metastasis to the lung or brain is the first manifestation of the disease[12].

The morphology of alveolar soft part sarcoma is very distinctive.It consists of circumscribed nests of large, round to oval eosinophilic cells, often showing central dyscohesion and hence an alveolar architecture. However, head and neck lesions in children more often have a solid growth pattern. The nests are surrounded by fibrous septa that often contain prominent vessels. The tumour cells themselves have copious, somewhat granular cytoplasm and eccentric rounded nucleoli with a prominent nucleolus. Although necrosis and hemorrhage may be prominent, nuclear pleomorphism and mitotic activity is very limited or scarce. Vascular invasion is a constant, striking finding that explains the tendency of the tumour to develop metastasis at an early stage of the disease.

In no more than 50% of the cases, periodic acid - Schiff - diastase staining reveals intracytoplasmic granules and crystalline rods, ultrastructurally which corresponds to membrane bound crystalline or filamentous material that seem to originate close to Golgi apparatus and that often adopts remarkable geometric shapes. These have been shown to consist principally of complexes of monocarboxylate transporter 1 and Cd147.

The histogenesis of this tumour has been argued ever since its description in 1952 and has not yet been definitely established. Interest has centred on a myogenic origin as a result of the finding of desmin, actin and other muscle markers. However, staining for desmin is usually only focal. A skeletal muscle origin is not supported by the detection of cytoplasmic MyoD1 protein because more specific MyoD1 or Myogenin nuclear staining is absent. Cytogenetic studies of this tumour have identified a specific alteration, der(17)t(X;17)(p11.2q25)[13]. This unbalanced translocation results in the fusion of the TFE3 gene on Xp11.2 to ASPSCR1 on 17q25. The resulting fusion gene encodes for a fusion protein that localizes to the nucleus and functions as an aberrant transcription factor[13,14]. The presence of this unique fusion gene also suggests that alveolar soft part sarcoma lacks a normal cellular counterpart.

CONCLUSION

No other sarcoma has quite the same appearance as Alveolar soft part sarcoma and ; however the alveolar appearance as well as microscopic picture of this entity can be closely simulated by other malignant neoplasms, viz. Renal cell carcinoma, Alveolar rhabdomyosarcoma, Malignant melanoma, paraganglioma, and granular cell tumour. Histomorphological features and immunohistochemistry helps in the definite diagnosis of these tumours.

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