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Case report

Retromolar monophasic synovial sarcoma: a rare entity in a rare location

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Abstract

Synovial sarcoma (SS) is a rare tumour that arises mainly in the peri articular tissues of the extremities. The majority of SS are biphasic on histology. Monophasic synovial sarcoma (MSS) of the mandible is a rare, aggressive malignancy having non-specific clinical and histological features with a tendency for distant metastasis. A 17- year-old girl presented to the surgical clinic with a gradually enlarging, painless ulcerated, right retro molar mass. Biopsy of the lesion revealed mitotically active, elongated, spindle cells arranged haphazardly and as fascicles with a hemangiopericytoma-like vascular pattern. The differential diagnosis included both odontogenic and non-odontogenic tumours. Immunohistochemistry showed positivity for AE1/AE3, CD99 and BCL2. CD34, CD31, SMA, CD117 and p63 were negative, thereby confirming a diagnosis of MSS. This case highlights key features of MSS and focuses on the importance of including it in the working differential diagnosis of malignant spindle cell tumours of the oral cavity.

Keywords: retro molar, monophasic, synovial sarcoma.

Introduction

Synovial sarcoma (SS) is a rare, soft tissue tumour that is thought to arise from pluripotent mesenchymal cells (1,2). It accounts for 5.6-10% of all soft tissue sarcomas (1,2). SS usually occurs in the para-articular regions of the extremities, mainly in relation to the joint capsules, tendon sheaths and bursae (1,2,3). Approximately 3 – 10 % of these tumours occur in the head and neck region([1,2), where the common sites of involvement include the hypopharynx, post pharyngeal region and para pharyngeal space (1,2,4,5,6). The occurrence of intra oral SS is extremely rare (4,6). Furthermore, intra oral SS lacks a definite clinical presentation (1) and has a diverse morphology thereby making its diagnosis challenging (1,4). We report a case of retromolar monophasic synovial sarcoma (MSS) occurring in a 17-year-old girl.

Case History

A 17-year-old- girl presented with a gradually enlarging, painless lump in the oral cavity of eight months duration. She was otherwise well with no co morbidities. On examination she had an ulcerated retromolar mass on the right lower jaw.

A biopsy of the mass showed a tumour comprising, elongated, spindle cells arranged haphazardly and as fascicles. The constituent cells had vesicular nuclei and scanty cytoplasm (Figure 1). Numerous blood vessels were present. Some had a hemangiopericytoma-like vascular pattern (Figure 2). Mitoses were 8 per 10 high power fields with atypical forms.

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Poorly differentiated areas and lympho-vascular invasion were not seen.

Immunohistochemistry showed positivity for AE1/AE3, CD99 and BCL2 and negativity for CD34, CD31, SMA, CD117 and p63, confirming a diagnosis of monophasic synovial sarcoma (Figure 1)

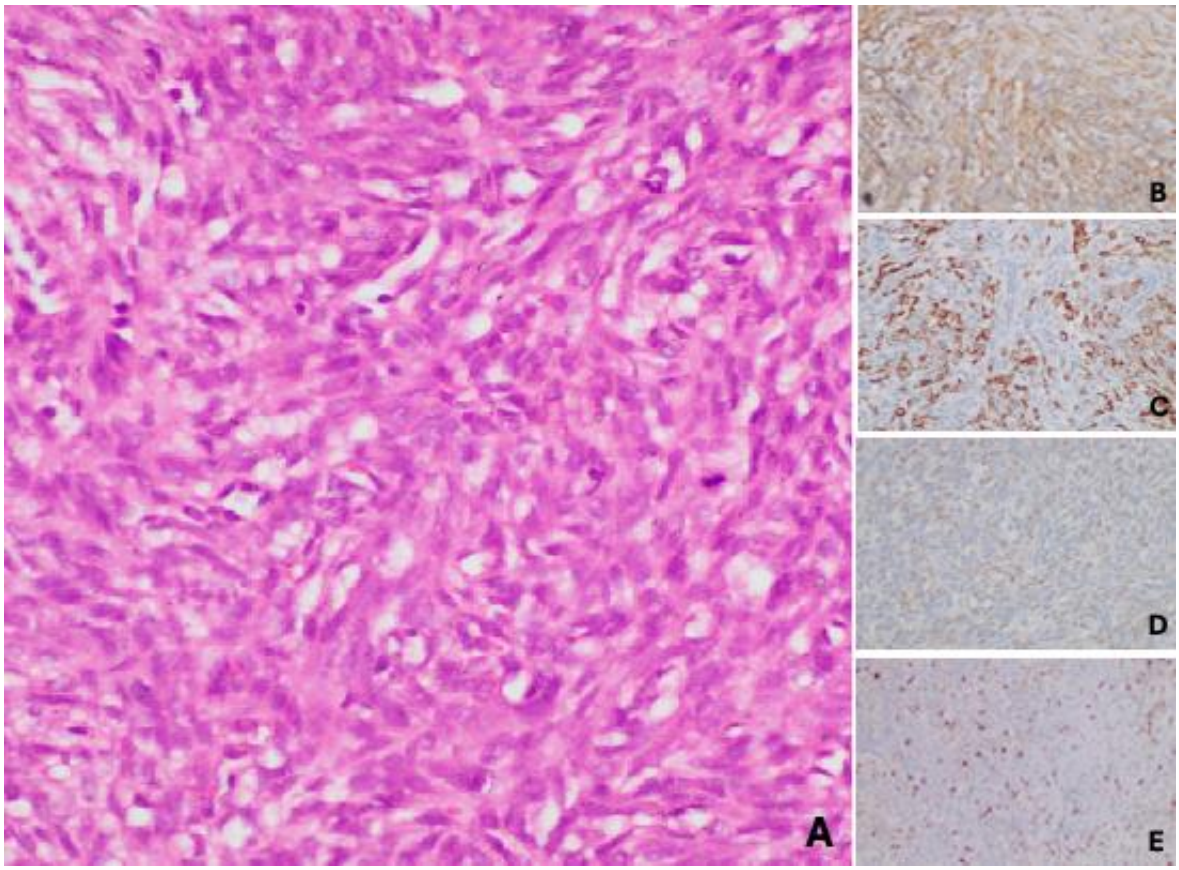

Figure 1: A) Hypercellular fascicles of spindle cells with ovoid to elongated vesicular nuclei, inconspicuous nucleoli and amphophilic cytoplasm - (Haematoxylin and eosin x400)B) The tumour cells showed cytoplasmic positivity for AE1/AE3 (AE1/AE3x 400); C) membrane positivity for CD99 (CD99x400), and D) cytoplasmic positivity for BCL-2 (BCL2x400). E) Ki67 highlighted the high proliferative activity of the tumour.

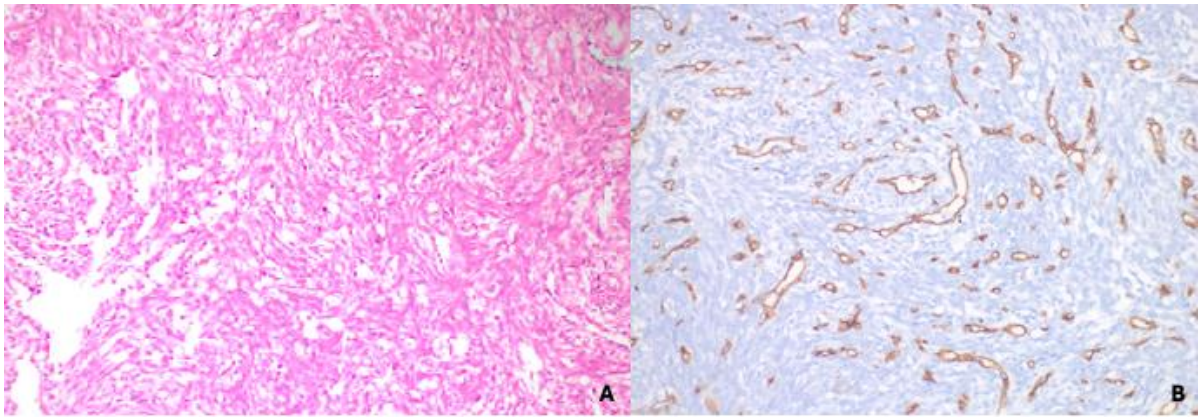

Figure 2: A) The tumour showed a haemangiopericytoma-like vascular pattern- (Haematoxylin and eosin x200) that was B) highlighted by immunohistochemistry for CD34 (CD34x400

Discussion

SS is an aggressive tumour of mesenchymal origin with a tendency for distant metastasis especially to lung, lymph nodes and bone marrow (2,7). It commonly occurs in young adults and has a slight male predominance (2). SS is a rare tumour that arises in the extremities in relation to the peri articular tissues. Head and neck SS accounts for 3 to 10% of tumours. The majority of head and neck SS occur in the hypopharynx, post pharyngeal region and para pharyngeal space. The occurrence of SS in the oral cavity is extremely rare.

SS is usually a slow growing tumour that gradually increases in size over 1 to 2 years (1). Patients with oral tumours may present with a swelling in the mouth, dysphagia, hoarseness or headache depending on the site of the original tumour and its spread (6).

The diagnosis of SS is based on its morphology, immunohistochemical findings and identification of specific translocations. Macroscopically, synovial sarcomas are well to poorly circumscribed tumours with a variegated appearance due to haemorrhage and necrosis (8). Histologically, SS is classified as either biphasic or monophasic (8). Either of these can have poorly differentiated areas (8).

Biphasic SS comprises varying proportions of epithelial and spindle shaped cells. The epithelial cells are cuboidal to columnar and have ovoid, vesicular nuclei and abundant pale eosinophilic cytoplasm (8). They are arranged as solid nests, cords or glands containing epithelial mucin (8). The spindle cell component is arranged as vague fascicles, dense sheets or in a herringbone pattern. The intervening stoma may be scanty or comprise thick bands of collagen (8). Areas of calcification or myxoid change may be present (8). The spindle shaped cells are small with scanty cytoplasm and ovoid, nuclei with granular chromatin and inconspicuous nucleoli (8). Monophasic tumours comprise spindle shaped cells of similar morphology to those seen in biphasic SS (8). Some tumours have a 'haemangiopericytoma-like' vascular pattern (8). Poorly differentiated areas are characterized by pleomorphic, spindled, round or epithelioid cells, thin fibrous septae, a mitotic count of more than 10 per 10 high power fields and necrosis (8).

Biphasic SS does not usually pose a diagnostic challenge. Monophasic SS, especially those that arise in atypical locations, on the other can be diagnostically challenging. The histopathological differential diagnosis considered for such a spindle cell lesion occurring in the oral cavity includes both non-odontogenic and odontogenic tumours. Non-odontontogenic tumours considered include leiomyosarcoma, monophasic (fibrous) synovial sarcoma, undifferentiated pleomorphic sarcoma, angiosarcoma, gastrointestinal stromal tumour (GIST), spindle cell rhabdomyosarcoma and fibrosarcoma. Odontogenic tumours in the differential diagnosis include odontogenic fibroma, ameloblastic fibroma and ameloblastic fibrosarcoma/ odontogenic sarcoma.

Odontogenic fibroma is a benign tumour comprising cellular fibro-collagenous tissue with inactive odontogenic epithelium arranged as scattered, small, strands, cords or nests (9). Ameloblastic fibroma is also a benign tumour comprising small islands and cords of attenuated ameloblastic epithelium within an immature, dense, collagenous stroma. (10) Ameloblastic fibrosarcoma/ odontogenic sarcoma is a biphasic tumour with benign odontogenic epithelium and a malignant stroma consisting of spindled cells showing pleomorphism, hyperchromasia, and increased mitoses (11). The lack of an epithelial component the present case favoured a non-odontogenic origin.

Immunohistochemical (IHC) stains help to differentiate monophasic synovial sarcoma from other non-odontogenic spindle cell lesions. Co-expression of mesenchymal markers such as vimentin and epithelial markers such as cytokeratin or EMA in the spindle cells is seen in SS but rarely expressed in other tumours. More than 90% of SS show focal expression of CK and EMA (12). This together with positivity for CD99 and BCL 2 and a negative staining for SMA, desmin, Myo D1 and S100, supports the diagnosis of SS. TLE1 is an IHC marker that has proved to be sensitive

in detecting synovial sarcoma. However, it can stain positive in other sarcomas and therefore is not specific and should be interpreted with caution (8, 13).

SS possesses the t(X;18)(p11;q11) translocation where the *SS18* gene on chromosome 18 is fused to *SSX1, SSX2* or *SSX4,* genes on chromosome X (8,14). This results in the formation of SS18-SSX fusion protein. This translocation is seen in 90% of the cases and can be detected with reverse transcription – polymerase chain reaction (RT-PCR) and fluorescent in situ hybridization (FISH) (8). IHC using antibodies to the SS18-SSX fusion-protein and the C terminus of SSX have been found to be highly sensitive and specific for SS and could replace molecular genetic or cytogenetic testing in most cases (14). Molecular testing is not required if the diagnosis of SS is possible using clinical features, histology and immunohistochemistry (4).

There are a number of clinico-pathological factors that determine the prognosis of SS. Poor prognostic factors include tumours with more than 20% of poorly differentiated areas, older age at diagnosis, tumour size ≥ 5 cm, non-extremity location, higher percentage of tumour necrosis, mitotic activity ≥ 10 per 10 high power fields or higher Ki67 activity, high tumour grade based on Fédération Nationale des Centres de Lutte Contre le Cancer (FNCLCC) method and positive surgical margins (8).

Due to the rarity of the tumour in the oral cavity, the optimal management protocol for oral synovial sarcoma has yet to be established (4). Wide local excision is the currently recommended treatment (1-7). This may be supplemented with adjuvant radiotherapy especially if the margins are positive (1-4). The role of chemotherapy in synovial sarcoma shows conflicting results (1,12). The plan of management for our patient was wide local excision with adjuvant radiotherapy. However, this was not possible as the patient was reluctant to undergo surgery and subsequently defaulted treatment

Conclusion

Monophasic synovial sarcoma of the mandible is a very rare, aggressive malignancy with a tendency for distant metastasis. In this case, the unusual location, vague clinical presentation, and non-specific histological features made diagnosis particularly challenging. This report aims to highlight key diagnostic features and emphasize the need to consider monophasic synovial sarcoma in the differential diagnosis of malignant spindle cell tumors in the oral cavity.

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