CASE REPORT

Periosteal desmoid tumour: a rare finding in the oral cavity

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SUMMARY

A 55-year-old female patient reported with an intraoral well-localised asymptomatic swelling on the right side of the mandible in relation to the right mandibular first molar along with a history of trauma 6 months back. Panoramic radiograph revealed normal trabecular bone pattern in relation to the lesion. The lesion was excised along with the associated buccal cortical plate and tooth. The microscopic examination revealed a well-circumscribed lesion consisting of spindle cells arranged in storiform pattern associated with the buccal cortical plate. The key feature to note was presence of reactive periosteum, which was in continuum with the lesion. Immunohistochemistry (IHC) revealed faint nuclear positivity for β -catenin. The above findings led to our diagnosis of periosteal desmoid, which is rarely reported in the head-and-neck region. The major challenge in the diagnosis of such lesions is good clinicopathological correlation as the differential diagnosis of spindle-cell lesions is vast and needs IHC confirmation.

BACKGROUND

Fibrous tumours represent a diverse subtype of soft-tissue tumours, and the spectrum can vary from benign conditions to intermediate malignancy to frankly malignant sarcomas. Fibromatoses are a heterogeneous group of distinct entities that differ in biological behaviour, but are histologically very similar. This group of fibrous tumours or tumour-like lesions poses significant difficulties in pathological diagnosis. They consist of proliferating well-differentiated fibroblasts, which may show local infiltrative pattern. Soft-tissue fibromatoses arising from musculoaponeurotic tissue, fascia or periosteum are termed as desmoid tumours. Around 26% of head-and-neck desmoids arise from the soft tissues including the periosteum around the mandible.^{1 2} The desmoids associated with intact periosteum are termed as periosteal desmoids. They can be well delimited, but majority of them are solitary firm mass with infiltrative borders.³

CASE PRESENTATION

A 55-year-old female patient reported to our institution with an intraoral swelling on the right side of the jaw, which was completely asymptomatic. The patient gave a history of trauma 6 months back. The patient had noticed a small swelling initially, which gradually progressed to the current size. Extraoral examination was unremarkable.

Clinical intraoral examination revealed a well-localised swelling in the right lower buccal vestibular region in relation to the right mandibular first molar (46), measuring approximately 1×1 cm. The overlying mucosa was stretched; however, no surface ulcerations were evident. The teeth associated (45, 46) were compromised periodontally and showed grade II mobility but were vital. On palpation, the swelling extended from the distal aspect of the mandibular right second premolar (45) to mesial aspect of second molar (47) and was associated with the buccal cortical plate (figure 1). It was painless and non-tender on palpation. The remainder of the head-and-neck examination was within normal limits with no significant lymph node enlargement in the submandibular and cervical region of the same and contralateral region.

Investigations

Orthopantomograph revealed no changes in the bone in relation to the lesion. The trabecular pattern of bone was normal, although 45 and 46 showed horizontal bone loss (figure 2). A faint hazy shadow was seen in the 46 region.

Under local anaesthesia, the lesion was surgically excised and submitted for histological evaluation with a surgical note that the lesion had to be removed along with a part of the buccal cortical plate (figure 3A) and tooth 46, as it was compromised periodontally.

Gross examination revealed a soft-tissue mass along with the associated buccal cortical plate measuring approximately 1.5×0.7 cm in size. The soft-tissue mass was well circumscribed, lobulated with smooth surface and was firm in consistency. The cut section appeared creamish white in colour and showed a lobular architecture (figure 3B).

Microscopic examination revealed a well-circumscribed fibrocellular lesion with cells arranged in lobular pattern with intervening connective tissue stroma. The lesional cells were spindle in shape running in short fascicles and in storiform pattern (figure 4A,B). The spindle-shaped cells appeared mature with predominantly ovoid nuclei. At areas, the cells were arranged in syncytium with no clear distinct cell borders (figure 5A,B). There was no mitosis, pleomorphism or atypical cells. Collagen arranged in the form of fibres and bundles associated with spindle cells were evident. Some of the fibres were wavy in nature resembling elastic fibres (figure 6). Endothelial lined blood vessels were evident with perivascular hyalinisation.

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Figure 1 Intraoral photograph showing a swelling in the right mandibular buccal vestibular region in relation to 46.

The decalcified section showed viable bone with compact cortical plate and fibrofatty marrow tissue. The important feature to note was the presence of reactive periosteum, which was in continuum with the lesion (figure 7A,B).

The histopathological findings confirmed the lesion to be benign as there was no evidence of any mitotic activity, necrosis or invasion into deeper tissues and bone. Both histopathologically and clinically, the lesion was well circumscribed. As the cells were spindle in morphology associated with collagen fibres, the lesion was suspected to be fibroblastic in origin. The radiographic detailing of the present case was normal while the surgical, grossing and histopathology showed the lesion to be arising from periosteum; hence, a final diagnosis of periosteal desmoid tumour was arrived at.

Immunohistochemistry was done and the cells showed faint nuclear positivity for β -catenin (figure 8).

Differential diagnosis

The differential diagnoses considered were traumatic fibroma, benign fibrous histiocytoma, collagenous fibroma and

Figure 2 Panoramic radiograph showing normal trabecular pattern of bone in relation to 45 and 46. The tooth was periodontally compromised with horizontal bone loss.

Figure 3 (A) Gross excisional specimen measuring approximately 1.5x0.7 cm with smooth lobular surface and creamish white colour. The arrow indicates attachment of bone to soft-tissue mass. (B) Cut section appears creamish white in colour with well-circumscribed lobular architecture.

desmoplastic fibroma. The mass was associated with reactive periosteum and was in continuum with the periosteum as could be co-related with the surgical note, grossing and histopathological findings. It could be thus concluded that the fibrous tumour arose from the periosteum and hence was diagnosed as periosteal desmoid tumour.

OUTCOME AND FOLLOW-UP

A regular follow-up of 12 months, though a short period, has reported no recurrence.

DISCUSSION

The group of soft-tissue tumours consisting of proliferating spindle cells admixed with collagen fibres showing absence of cytological and clinical malignant features are grouped under the term fibromatosis. It is microscopically benign but can be locally invasive with a tendency to recur.⁴

The soft-tissue fibromatoses arising from musculoaponeurosis, fascia or periosteum are termed as desmoid tumours and constitute a rare fibroblastic proliferative disease. The term desmoid is derived from the Greek word '*desmoids*', which means *band-like*. The soft-tissue masses are fixed to the surrounding tissue and are firm in consistency. They are slow-growing, histologically benign tumours. The lesions are locally aggressive and invade the surrounding structures though they appear benign.^{2,5}

The clinical course of the disease is unpredictable, but various studies have suggested that there is an initial phase of growth, followed by a period of growth arrest. The tumour may stabilise over time or even regress.⁵

The incidence is rare accounting for 0.03% of all neoplasms and is estimated at 2.4 to 4.5 cases per million people annually. Desmoid tumours are grouped into intra-abdominal, abdominal and extra-abdominal types based on their location. Shoulder

Figure 4 (A) Photomicrograph showing a lesion consisting of spindle-shaped cells in short fascicles and storiform pattern (H&E, x40). (B) Cells arranged in nodular pattern with intervening connective tissue (H&E, x40).

Figure 5 (A) Spindle-shaped cells appear mature with predominantly ovoid nuclei (H&E, $\times 200$). (B) Areas of cells arranged in syncytium with no clear distinct borders are seen (H&E, $\times 200$).

girdle, chest wall and inguinal regions are the most common extra-abdominal sites of presentation.^{5,6}

Around 7% to 15% of extra-abdominal fibromatosis are seen in the head-and-neck region, out of which 26% arise from the soft tissues, including the periosteum.¹ The most common locations in the head-and-neck region include neck (40%–85%), face, oral cavity, scalp, paranasal sinuses and orbital region.⁴ One tumour involved the tongue in a 24-case series reported in the head-and-neck region.⁷ The age of presentation ranges from 15 to 60 years with a mean age of 30 years at diagnosis. Desmoid tumours present with female predominance, the ratio being 2:1.^{6,8}

The well-delimited desmoids associated with intact periosteum are termed as periosteal desmoids, but majority of them are solitary firm mass with infiltrative borders. The lesion most commonly occurs in the extremities, the specific site being the posterior femur, and one case has been reported in the distal tibia.³

The present case was reported in the mandibular right molar region associated with the buccal cortical plate of a 55-year-old female patient. The lesion was asymptomatic but was an indolent progressive localised swelling.

Desmoid tumours can occur sporadically or as a manifestation of familial adenomatous polyposis (FAP), which is a hereditary autosomal dominant syndrome caused by germline mutations of adenomatous polyposis coli gene. If left untreated, FAP, a familial cancer predisposition syndrome, most of the time presents a marked tendency towards colorectal cancer at an early

Figure 6 Collagen is arranged in the form of fibres and bundles with spindle cells. Fibres are wavy in nature resembling elastic fibres (H&E, $\times 200$).

Figure 7 (A) Photomicrograph showing viable bone with compact cortical plate and fibrofatty marrow tissue (H&E, $\times 100$). Inset: high-power view of the lesion showing the periosteal reaction (H&E, $\times 200$). (B) Presence of reactive periosteum with cells arranged in syncytium in continuum with the lesion (H&E, $\times 100$).

age. In their lifetime, around 32% of patients with FAP develop desmoid tumours.^{5,6}

For majority of individuals affected, these tumours occur in a sporadic manner, not caused by any predisposing genetic disease. Such patients have no other health problems associated with mutations in the APC gene and have no close family members with the tumours. Chronic irritation or trauma, including any surgical trauma to the body parts, has been suggested to increase the risk of desmoid tumour.⁶ According to Goellner and Soule, immature fibroblasts in the region of the trauma lead to an uncontrolled attempt at healing and lead to tumour formation.⁴

The precise aetiopathogenesis of desmoid tumours is obscure. Some of the aetiological factors considered include dysregulation of connective tissue growth and involvement of hormone oestrogen. It has been observed that there is increased growth rate with high levels of the oestrogen hormone and regression of the tumour under conditions when the levels are decreased (like before menstruation, after menopause, after irradiation to ovaries and after administration of anti-oestrogen agent tamoxifen). The high expression of nuclear β -catenin, which is responsible for cell proliferation and survival both in sporadic and FAP-associated desmoid tumours, is characteristic. Inactivation of APC gene leading to accumulation of β -catenin is observed in FAP-associated desmoid tumours whereas mutation of CTNNB1 (gene coding for β -catenin) in 85% of cases is responsible for sporadic conditions. Nuclear positivity for β -catenin is relatively

Figure 8 Spindle cells showing faint nuclear positivity for β -catenin (immunohistochemical stain, $\times 200$).

high and helps to distinguish desmoid tumour from other histological mimics.⁴⁻⁸

The patient of the present case had no other clinicopathological findings and was not associated with APC gene mutations and hence was considered to be a sporadic case. The patient does give a history of trauma 6 months back and the age of the patient correlates with the menopausal stage.

Plain radiographs of extra-abdominal desmoid tumours are usually normal. Radiographs occasionally reveal a soft-tissue shadow when it is large or displaces adjacent soft-tissue structures, but calcifications are rare. Bone involvement by desmoid tumours can manifest as pressure erosions, periosteal reaction or cortical thickening.⁵ The orthopantomograph of the present case revealed no bone destruction in relation to the lesion, and the trabecular pattern of the bone at the site of lesion was well maintained.

Gross surface appears fibrous, well circumscribed, grey pink to white, firm mass associated with the periosteum in case of a periosteal desmoid. Sometimes the mass has an intracortical compartment in connection to the periosteal mass appearing dumbbell in shape.³ The present case showed a lobulated mass that was attached to the buccal cortical plate. The cut surface was fibrous, off-white in colour with lobulations.

Microscopically, desmoid fibromatoses constitute interlacing fascicles of elongated but relatively uniform spindle-shaped fibroblasts. The tumours are often poorly circumscribed and infiltrate into the adjacent soft tissue, but sometimes can be delimited. Spindle cells are uniform, pale eosinophilic with tapering or ovoid vesicular nuclei. Nucleoli are inconspicuous, with absence of nuclear atypia and hyperchromasia. Cellularity and mitotic activity are variable, but mitoses are generally sparse and do not hold much significance for prediction of the outcome. Cellularity can range from very sparse and fibrotic to relatively cellular, appearing almost storiform in areas.^{3,5}

The differential diagnoses considered for the present case were traumatic fibroma, benign fibrous histiocytoma, collagenous fibroma and desmoplastic fibroma. Considering the population of spindle-shaped cells without any inflammatory reaction, traumatic fibroma can be ruled out as it appears as a nodular mass of connective tissue, which is dense, collagenised, shows inflammatory cells and is lined by stratified squamous epithelium that can be normal or atrophied.⁹

Collagenous fibroma appears as a hypocellular lesion, composed of fibroblastic/myofibroblastic cells dispersed in a collagenous-rich stroma. The cells vary from spindle to stellate shape and are often binucleated with large elongated to oval nuclei. Nuclei exhibiting delicate chromatin and small eosinophilic nucleoli may be present whereas in case of desmoids the spindle cells are uniform and nucleoli is inconspicuous, similar to our case.¹⁰

The storiform pattern of spindle cells is classic of benign fibrous histiocytoma, but other features such as round histiocyte-like cells, xanthoma cells or myxoid areas were not evident in our case, thus narrowing our diagnosis.⁹

Lastly, as the histopathology exactly mimics desmoplastic fibroma (DF), it was difficult to differentiate between the two lesions, but DF is always intraosseous.¹ The present lesion was associated with reactive periosteum distinguishing it from DF.

Taking into account the clinical presentation of the lesion, paraosteal osteosarcoma can also be considered as a differential diagnosis, but histopathology clearly ruled out paraosteal osteosarcoma as the present lesion lacked tumour osteoid, bony invasion, pleomorphism and other classical features of malignancy. Thus, the clinical presentation, gross features presenting with

attachment to buccal cortical bone, histological evidence of a benign fibroblastic proliferation with variable collagen, elastic fibres and periosteal reaction pointed the lesion to be called as periosteal desmoid tumour.

Treatment of desmoid tumour mainly consists of surgical wide excision, with or without adjuvant radiotherapy. Primary radiotherapy and systemic approach is advocated for inoperable/unapproachable areas to avoid surgical morbidity. Systemic approach includes chemotherapy (imatinib), use of non-steroidal anti-inflammatory drugs and hormones. However, systemic treatment for head-and-neck desmoid tumours has been used infrequently. In few unresectable cases where local treatment leads to high morbidity, a protocol of watchful waiting until significant symptoms develop may be followed as these lesions tend to have periods of stability or may even regress.⁹ Since the lesion was relatively small in size, a wide surgical excision was planned for the current case.

The local recurrence of desmoid tumours in the head-and-neck region after surgery, which may or may not be followed by radiotherapy, in a 40-year literature review is 30%. Previous studies have reported a recurrence range of 43% to 70%. A recurrence rate of 21% in sinonasal region and 20% in oral cavity desmoid tumour is reported.⁹

CONCLUSION

Periosteal desmoids are very rare in the gnathic skeleton and histologically overlap DFs. Though cases of soft-tissue desmoids have been reported, cases arising or associated with the periosteum of the mandible have been rarely reported. The present case highlights the importance of thorough clinicopathological correlation, good histopathological sampling and observation to arrive at the diagnosis. Though benign, these lesions require recall on a regular basis.

Learning points

- Periosteal reactions are seen in elderly patients also, unlike assumed to be seen in only adolescent and young individuals.
- Periosteal reactive lesions should be ruled out before diagnosing periosteal malignancies like periosteal osteosarcoma.
- Lesions attached to the bone need to be evaluated as periosteal bone lesions despite their fibrous nature.
- A diagnosis of desmoid tumour should be considered in cases of well-delimited monotonous spindle-cell proliferations in the absence of inflammatory cells.
- A good clinical, radiographical correlation and histopathological sampling is essential in understanding the origin of the tumour. Many times, orientation of the lesion and histopathological correlations are the clues to arrive at the diagnosis. In our case, an association with the buccal periosteum of the mandible helped us in arriving at the diagnosis.

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