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CASE REPORT OPEN ACCESS

A case report of desmoplastic fibroma in a three-year-old black African girl

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Abstract
Background: Desmoplastic fibroma is a very rare bone tumour that is commonly recognised by its striking resemblance to desmoid fibromatosis, commonly seen in soft tissues.
Case Presentation: A 3-year-old girl presented to the Oral and Maxillofacial Department with swelling on the lower jaw for 4 months duration. Extra-oral examination revealed: Facial asymmetry due to the discrete swelling on the right mandible measuring 14 x 12cm in widest diameter below the right pre-auricular region to the right mental region. The Craniofacial CT scan showed an expansile jaw mass with destruction of the mandible. There was no evidence of bone formation. Histopathology diagnosis was desmoplastic fibroma of the bone. The tumour cells were weakly positive for MyoD1.
Conclusion: Pathologists must know that cells of desmoplastic fibroma may sometimes be weakly positive for MyoD1, as in rhabdomyosarcoma.

Keywords: Desmoplastic fibroma, Mandible, Immunohistochemistry, Biopsy, Spindle cell

Background

Desmoplastic fibroma is a very rare bone tumour that is commonly recognised by its striking resemblance to desmoid fibromatosis, commonly seen in soft tissues (1). It is mostly found in children and adolescents. It is mostly situated in the mandible (2). It is quite similar in histologic architecture to low-grade central fibroblastic osteosarcoma but can sometimes be differentiated from it based on the absence of osteoid formation (3). We present a rare case of desmoplastic fibroma of the mandible seen at our centre.

Case presentation

A 3-year-old girl presented to the Oral and Maxillofacial Department with swelling on the lower jaw lasting 4 months. The mother was said to have initially felt a small, hard swelling while attempting to clean her daughter's face in the morning. The swelling was initially about 2cm and painless but has progressively increased in size. There is associated difficulty in masticating and swallowing. There is no difficulty with breathing and no history of weight loss. There was no history of trauma to the jaw, discharge from the swelling, or change in speech. There was no history of exposure to radiation or chemicals. There is no family history of this condition. Extra-oral examination revealed facial asymmetry due to the discrete swelling on the right mandible, measuring 14 x 12cm in widest diameter below the right pre-auricular region to the right mental region. The mass was bone-hard, slightly tender, and attached to the underlying structure. The overlying skin was intact, with no differential warmth. Level II nodes (ipsilateral) and level Ib nodes (contralateral) are palpable, discrete, firm, and freely mobile. Intra-orally, there was no limitation in mouth opening and no halitosis.

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There was fair oral hygiene. The lingual extent was, however, less in the ipsilateral region. The overlying mucosa was intact. The craniofacial CT scan showed an expansile jaw mass with the destruction of the mandible (Figure 1). There was no evidence of bone formation. She was subsequently worked up for an incisional biopsy under general anaesthesia, and the tissue sample removed was sent for histopathologic examination and immunohistochemistry. The histologic section showed spindle and stellate cells in a swirling pattern. There were few `small-calibre vascular channels. There was no osteoid formation or permeation of the inter-trabecular space of the bone. This is shown in Figure 2. A few of the cells were positive for S100(Figure 3) and MyoD1(Figure 4) but negative for pancytokeratin and desmin. The spindle cells in the index case were negative for SMA. We could not stain for MSA and caldesmon. Histopathology diagnosis was desmoplastic fibroma of the bone. There was no post-incisional complication. After the diagnosis, the mass was resected with margins free of tumours. The patient was without recurrence when seen three months after surgery.

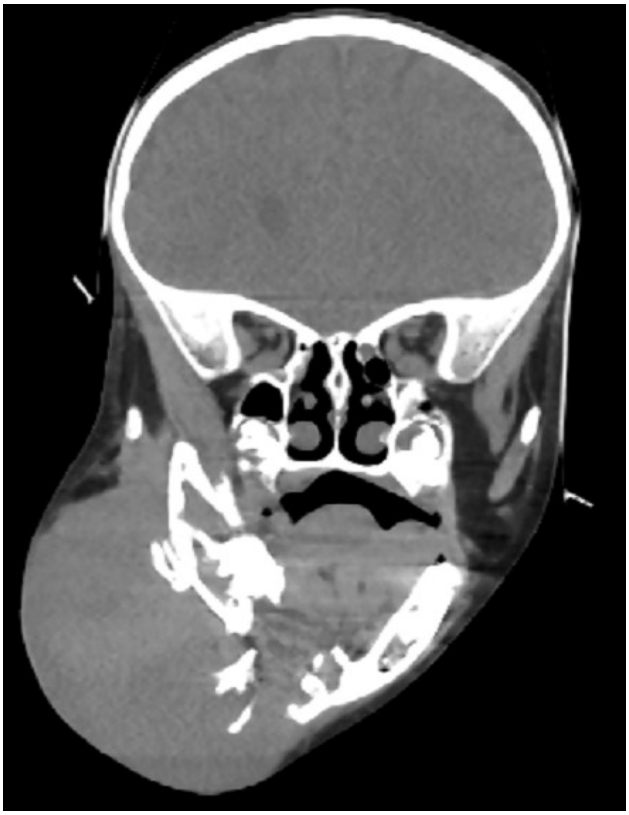

Figure 1 Craniofacial CT-Scan

The CT scan shows a right-sided mandibular mass. We found the widest diameter to be 10cm. There is the destruction of the mandible and infiltration of the soft tissue by the tumour.

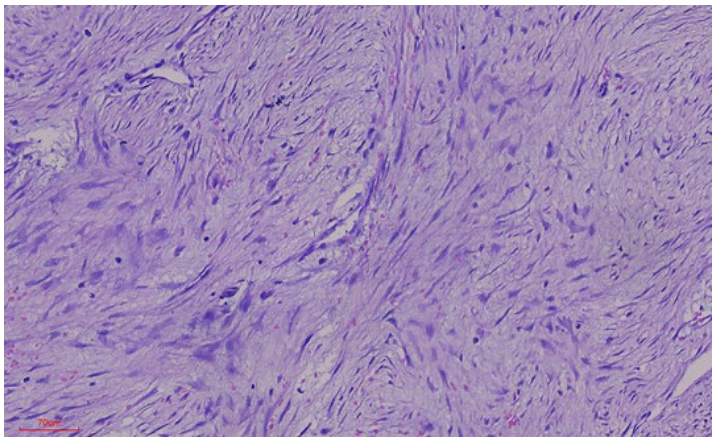

Figure 2: Haematoxylin and eosin-stained microscopic section of the mass

The photomicrograph shows spindled and stellate cells in a swirling pattern. There are few small calibre vascular channels. There was no osteoid formation or permeation of the inter-trabecular space of the bone.

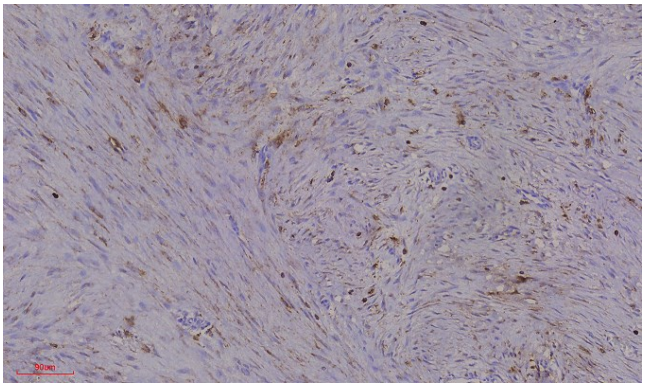

Figure 3: S100 immunohistochemistry

There is weak staining of many of the neoplastic cells by the S100 antibody.

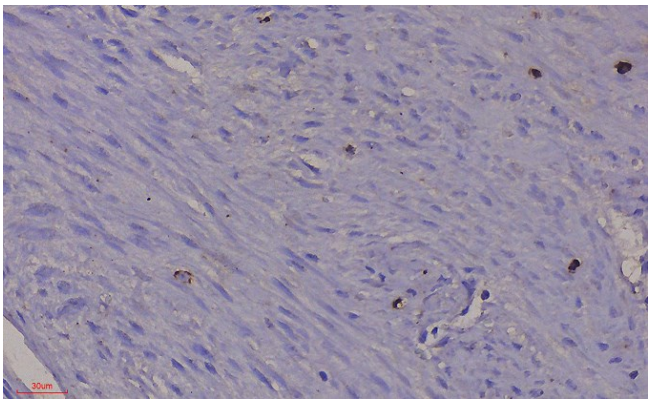

Figure 4: MyoD1 immunohistochemistry

Few of the cells are stained by MyoD1.

Discussion

Our index case highlights a maxillofacial bone tumour. The diagnosis of this group of tumours is known to be challenging (4). The various differential diagnoses and the varying treatment modalities necessitate reaching a definitive diagnosis in every case. Although this can be difficult in a resource-limited setting like ours, utilizing clinical and available imaging information can be very useful (5, 6).

Many authors have reported that desmoplastic fibroma of the bone can be seen in very young patients (7, 8, 9, 10). Hence, the index case being a three-year-old is consistent with reports from various authors. Pathologists should have a high index of suspicion for this tumour when the histology of a bone lesion shows sheets of bland spindle-shaped cells devoid of bone matrix, especially in an appropriate radiologic context.

Desmoplastic fibroma of the bone is an extremely rare tumour that can easily be identified on hematoxylin and eosin-stained sections by its striking resemblance to desmoid fibromatosis of soft tissues. Some authors have reported its occurrence in jaw bones, as was seen in the index case (5, 11, 12). Although it can be suspected in routine microscopy, it is always important to distinguish it from morphologically similar lesions, most of which are more common. Fibroblastic osteosarcoma is a more common lesion that can mimic desmoplastic fibroma of the bone. Fibroblastic osteosarcoma is more commonly found in the distal femur, proximal tibia, and proximal humerus than other conventional osteosarcomas. It is less likely to be found in the mandible. However, since desmoplastic fibroma can be found in long bones, it becomes an important differential diagnosis in spindle cell tumours in such locations (13, 14, 15). Magnetic resonance imaging (MRI) and CT scans can identify osteoid formation in fibroblastic osteosarcomas. There is typically no osteoid formation in the desmoplastic fibroma of the bone. Similarly, osteoid matrix, or

woven bone, is commonly seen in fibroblastic osteosarcoma but not desmoplastic fibroma of the bone. The nuclei of spindle cells in fibroblastic osteosarcoma are commonly atypical and not bland, as seen in the index case.

Another major differential diagnosis in our index case is low-grade central fibroblastic osteosarcoma. This tumour is made up of proliferating spindle-shaped cells that usually look bland, like the spindle cells in the index case. MRI can be very useful in identifying bone formation that is typical of these cases. An osteoid matrix can also be identified on hematoxylin and eosin-stained sections. It is also important to note that it is more common in bones where other conventional osteosarcomas are found. In centres that have the required resources, genetic analysis is very important in distinguishing between low-grade central fibroblastic osteosarcomas and desmoplastic fibroma (16).

Due to the bland nature of the nuclei of the spindle-shaped cells, it is very important to consider other benign lesions as possible diagnoses. Benign tumours such as leiomyoma and neurofibroma should be considered. However, the spindle cells of Leiomyoma usually have more abundant pink cytoplasm than those of the cells seen in the index case and are usually positive for smooth muscle actin (SMA), muscle-specific actin (MSA), and caldesmon. The spindle cells in the index case were negative for SMA. We could not stain for MSA and caldesmon. Although some cells were stained for S100, as is seen in neurofibroma, the lesion is unlikely to be a neurofibroma due to the rapid growth and infiltrative attribute.

Some of the lesional cells were weakly positive for MyoD1. This is not usually reported in the literature. Pathologists must know that cells of desmoplastic fibroma can sometimes be weakly positive for MyoD1, as in rhabdomyosarcoma. Hence, spindle cell rhabdomyosarcoma may be considered a differential diagnosis. However, neoplastic cells in spindle cell rhabdomyosarcoma are strongly and diffusely positive for MyoD1 and do not form collagen, as seen in the index case. The nuclei in spindle cell rhabdomyosarcoma usually show severe atypia.

Conclusion

Pathologists should have a high index of suspicion for desmoplastic fibroma when the histology of a bone lesion shows sheets of bland spindle-shaped cells devoid of bone matrix, especially in an appropriate clinical and radiologic context. Due to the bland nature of the nuclei of the spindle-shaped cells, it is very important to consider other benign lesions as possible diagnoses. Pathologists must know that cells of desmoplastic fibroma may sometimes be weakly positive for MyoD1, as in rhabdomyosarcoma.

List of Abbreviation

CT: Computed Tomography
SMA: Smooth muscle actin
MSA: Muscle specific actin
MRI: Magnetic Resonance Imaging

Declarations

Ethical approval and consent to participate

Written informed consent was obtained from the patient's parent for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor.

Consent for publication

All authors gave consent for publication of the work under the Creative Commons Attribution-Non-Commercial 4.0 license.

Availability of data and materials

The tissue blocks and all essential data supporting the findings of this case are available upon reasonable request from the corresponding author.

Competing interests

The authors declare no conflict of interest.

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Authors' contributions

OOO and OCC reported the clinical and pathologic findings. OOO, OCC, UGC, OOZ and BOA contributed to the discussion and the final write up.

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