Peer Reviewed **Case Report**

THE ROLE OF MEDICAL IMAGING IN THE DIAGNOSIS OF DESMOPLASTIC FIBROMAS

Gerhardus GV Koch *MHSc: Radiography (D); PGDip: Tertiary Education; Professional Certificate: Computerised Tomography*

Faculty of Health and Wellness Sciences, Department of Medical Imaging and Therapeutic Sciences, Cape Peninsula University of Technology

https://doi.org/10.54450/saradio.2023.61.1.551

Abstract

A desmoplastic fibroma (DF) is a rare, fibroblastic tumour. DFs are known to be locally aggressive and challenging to diagnose. The role of medical imaging in diagnosing maxillofacial tumours is undisputable. In this case report a DF diagnosed in the mandible and surrounding soft tissue of a five-year-old female patient is presented. The patient's clinical history, radiological findings and management are discussed as well as the epidemiology, aetiology and treatment options.

Keywords. Medical imaging, maxillofacial, rare tumour

Lay abstract

A rare tumour diagnosed in a five-year-old female patient that affects her lower jaw and surrounding soft tissue is discussed.

CASE REPORT

A five-year-old female was referred by a local community health centre to the outpatient maxillofacial clinic at a tertiary hospital due to a progressively growing right-sided facial mass. No history of trauma was noted. On examination, a right-sided facial mass was observed, contributing to facial asymmetry, hard swelling and discomfort. The systemic clinical examination appeared to be within normal limits and the attending doctors discussed the patient with the radiology department for further investigation.

An orthopantomogram (i.e., panorex) of the maxilla and mandible was performed as the first imaging technique. This was followed by a biopsy of the mass lesion. The histopathological report anticipated a central myofibroma, however, the orthopantomogram revealed an aggressive lesion with periosteal reaction. No conclusive diagnosis could be made from this imaging examination. Due to the complexity of this case, the rare pathological occurrence, and inconclusive diagnosis, the following imaging examinations and pathological investigation were subsequently performed: 1) a computed tomography (CT) scan, 2) a larger representative biopsy sample including soft tissue, 3) a magnetic resonance imaging (MRI) scan, and 4) a nuclear medicine bone scan.

An unenhanced CT scan of the mandible was performed on a Philips Ingenuity Core 128 slice CT scanner at 5mm intervals. The raw data obtained were reconstructed to bony and soft tissue windows in the coronal, axial and sagittal planes as well as a three-dimensional reconstruction of the mandible with the aid of volume rendering. On the CT a hypodense soft tissue mass extending from the right temporal fossa to the right submandibular space, measuring approximately 79mm x 62mm x 64mm (Figures 1 to 3) was noted. Bony destruction and lytic changes were visible on the patient's mandible and zygomatic arch, symbolic of an aggressive growth pattern (Figure 4). No intracranial involvement was noted.

A second biopsy, to obtain a larger sample, was performed five days after the CT scan. The findings from the histopathological investigation indicated that the tissue sample consisted of benign spindle-shaped cells, vesicular nuclei, ordinary nucleoli and moderate amounts of eosinophilic cytoplasm in a variable collagenous background. No malignant features were evident.

Seven days after the second biopsy was performed, the patient underwent a soft tissue MRI scan of the head and neck to further evaluate the nature of the pathology. On the unenhanced T1-weighted images, the mass appeared isointense (similar to the appearance of normal muscle tissue) and hyperintense on the T2-weighted images (indicative of a pathological condition). Six days after the MRI scan, a nuclear medicine (NM) bone scan was performed; a significant

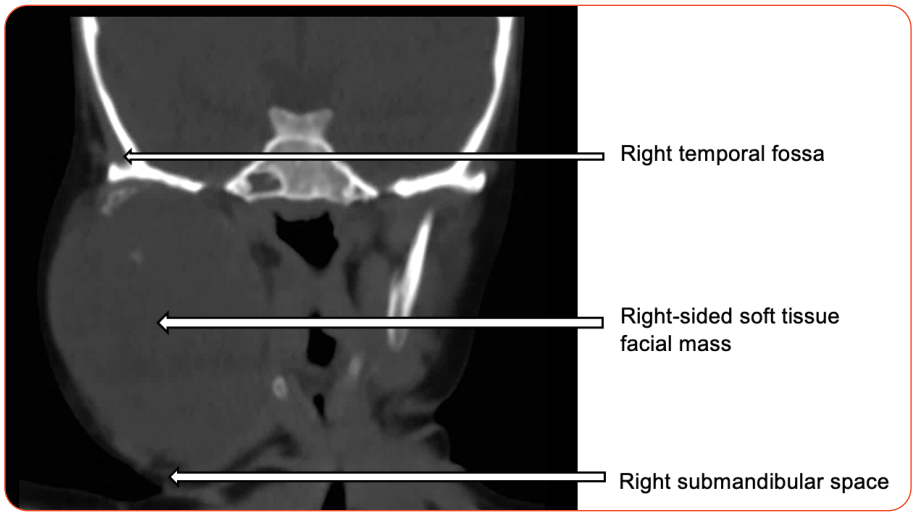

Figure 1. A coronal CT image demonstrating the position of the right-sided soft tissue facial mass: right temporal fossa to the right submandibular space.

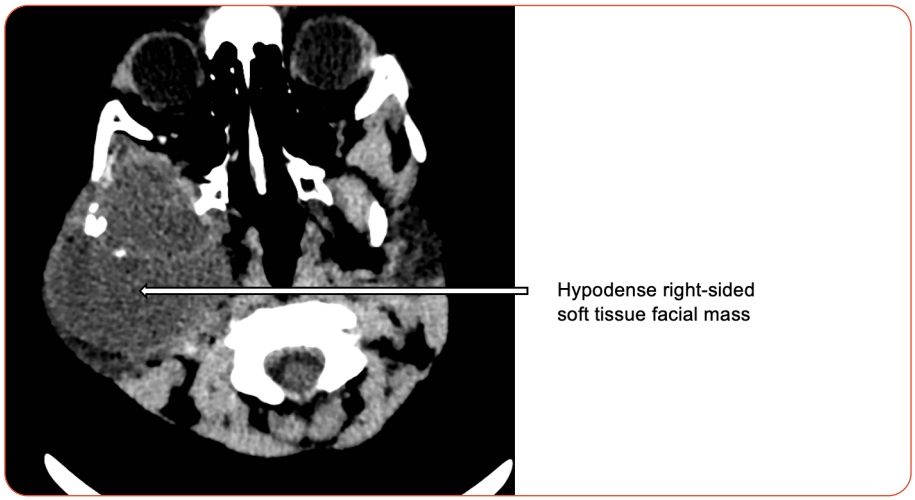

Figure 2. An axial CT image demonstrating the hypodense right-sided soft tissue facial mass.

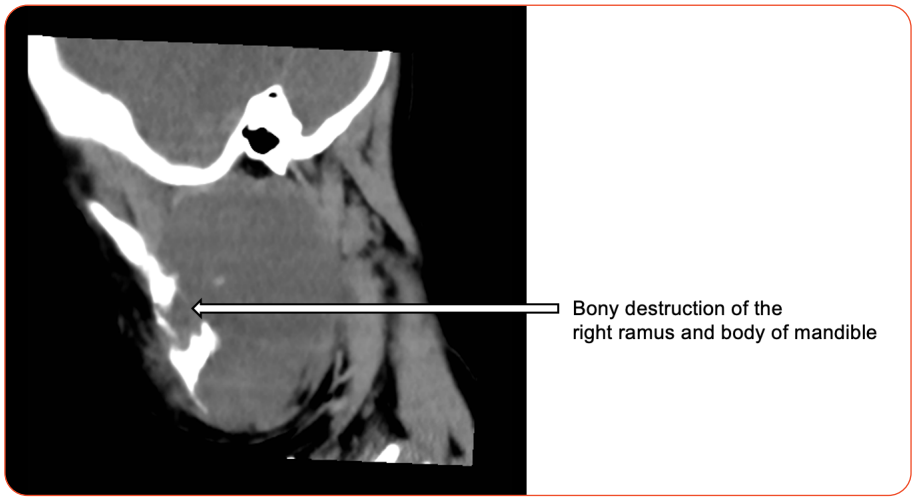

Figure 3. A sagittal CT image demonstrating the bony destruction of the right mandible.

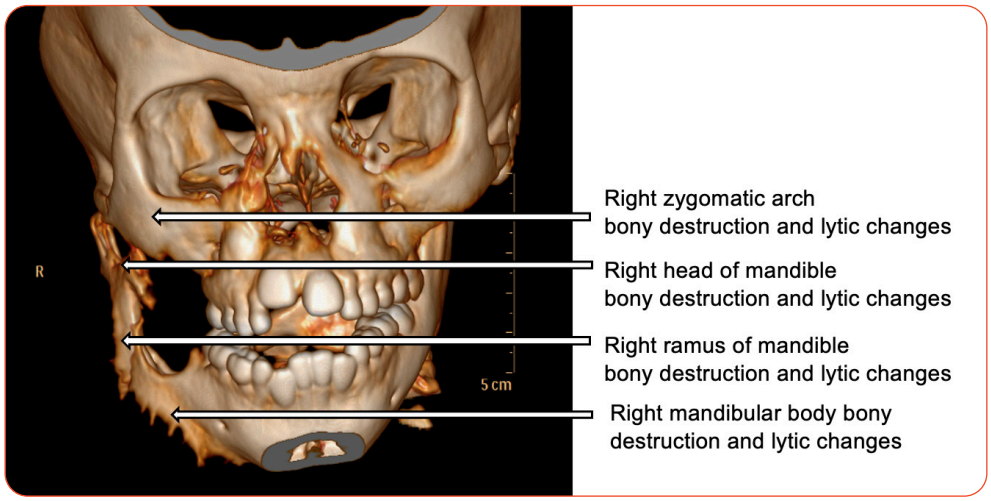

Figure 4. A three-dimensional CT image demonstrating bony destruction and lytic changes.

uptake of the radioactive tracer within most parts of the right mandible, maxilla, zygomatic arch, base of skull and orbital area was noted. This uptake of the radioactive tracer is indicative of a pathological condition.

Based on the findings from the second biopsy and additional imaging (CT and MRI scans), the patient was diagnosed with a primary, intraosseous DF. The diagnosis was further described as having both intraosseous and extraosseous involvement.

DISCUSSION

DFs affect approximately 0.11% of the population.[1] According to Averna et al,[2] DFs of the bone was first described in 1958. A DF can be defined as a rare, fibroblastic tumour, known to be locally aggressive and diagnostically challenging.[3] Histologically, DFs are difficult to diagnose due to its rare occurrence[2] and it being one of the most uncommon bone disorders.[1] There are only a few case reports available from radiology, orthopaedic and pathology literature.[4]

The patient presented in this case report was five years old and had a combination of intraosseous and extraosseous tumour involvement. A DF in the head and neck area can be classified either as a) intraosseous (in the bone), b) extraosseous (in the soft tissue), or c) a combination of both: intraosseous with an extraosseous extension.[5] Trauma and genetic factors are believed to be the cause of a DF; however, the exact aetiology remains unknown.[3,6,7] There is no prevalence associated with biological sex and from those cases reported in literature, a DF generally presents in patients younger than 30 years of age.[1,2,5,6,8,9]

Signs and symptoms associated with a DF of the mandible and surrounding soft tissue may include painful and localised swelling, dysphagia, facial asymmetry, abnormal positions of teeth and a loss of teeth.[1,2,3] Facial asymmetry, as well as painful and localised swelling is in keeping with the signs and symptoms of the patient presented in this case report.

For the diagnostic assessment of a DF, medical imaging plays a critical role. Imaging techniques such as planar imaging (i.e., general X-rays), CT, MRI and ultrasound have been reported on in literature for their unique contributions. General X-rays and CT are considered more accurate for the assessment of bony destruction.[9] Bony lesions associated with DFs are often described as having a "soap-bubbles" appearance; due to a thinned cortex and bony trabeculae's display as being lobulated.[9] MRI is best used for detecting soft tissue extensions related to DFs.[9]

According to Guruprasad and Chauhan,[5] CT imaging can be utilised to evaluate both the bone and soft tissue components of a DF. In bone, the typical appearances may include radiolucent, mixed radiolucent or mildly sclerotic matrix patterns.[8] An intraosseous DF of the mandible is often also associated with bony destruction and lytic changes of the head, ramus and body of the mandible.[7] Similar patterns were noted at CT for the patient presented in this case report (Figure 4). Diagnosing a DF on CT alone can be clinically challenging as many other tumours may also resemble similar radiographic features.[1]

A similar case report discussed findings of a colour Doppler ultrasound examination. Important information related to the soft tissue mass's morphology could be determined: solid, highly vascular mass with no haemorrhagic areas.[1] Another case report that discussed the role of ultrasound imaging and elastography in the diagnosis of a DF demonstrated the intermediate stiffness of the extraosseous region of the tumour. Enlarged laterocervical and submandibular lymph nodes were also identified and a surgical biopsy was performed on these nodes. A combination of imaging and a histopathological investigation allowed for an accurate diagnosis to be made.[10] When compared to the patient presented in the current case report, an ultrasound examination was not performed. A histopathological investigation, however, was performed in conjunction with a CT and MRI to confirm the five-year-old female patient's diagnosis.

A retrospective study conducted in 2014 on previously treated patients, found that a statistically significant relationship exists between the presence of extraosseous soft tissue extensions and the local recurrence of a DF.[11] This was supported by more papers that indicated a local recurrence rate of 37-72%, if not adequately treated.[1,8] Treatment options vary and is based purely on the size and extent of the tumour.[6] According to literature, the following options may be considered for the treatment of a DF: 1) radiotherapy, 2) chemotherapy, and 3) surgery.[6]

Radiotherapy, however, is not often recommended due to the low success rate and the increased likelihood for the manifestation of secondary, radiation-induced malignancies.[6] An aggressive surgical approach whereby the affected tissue is completely removed (including a 2-3cm margin adjacent to the affected tissue) is recommended.[11] An aggressive surgical approach increases the prognosis and success rate of a patient diagnosed with a DF.[11]

CONCLUSION

This case report presented a five-year-old female diagnosed with a rare, locally aggressive, DF of the mandible with soft tissue extension. Although a DF is known to be challenging to diagnose, it is evident that histopathology, together with the use of multiple imaging modalities play a vital role in the diagnosis of maxillofacial tumours. Different imaging modalities have unique strengths which should be exploited to ensure that the most appropriate diagnosis can be made in a timely manner. However, given that DFs affect young patients particular attention should be afforded to keeping the radiation dose to patients to a minimum to mitigate radiation-induced side effects.

CONFLICT OF INTEREST

None to declare.

ETHICAL CONSIDERATIONS

Permission for presenting this case report was obtained from hospital management. No identifiable details of the patient, healthcare professionals or healthcare institution have been revealed.

AUTHOR CONTRIBUTION

Sole authorship.

REFERENCES

1. Nedopil A, Raab P, Rudert M. Desmoplastic fibroma: a case report with three years of clinical and radiographic observation and review of the literature. The Open Orthopaedics Journal. 2013; 7(1): 40-46. [cited 2023 March 15]. Available from: https://doi.org/10.2174/1874325001307010040
2. Averna R, De Filippo M, Ferrari S, Bacchini E, Rossi C. Desmoplastic fibroma of the mandible. Acta Biomedica. 2011; 82(1): 77-81. [cited 2022 December 03]. Available from: http://www.mattioli1885journals.com/index.php/actabiomedica/paper/view/1369/998.
3. Mohamed S, Faical C, Mohammed M. Desmoplastic fibroma of the mandible. Open Access Journal of Translational Medicine & Research. 2017; 1(1): 20-22. [cited 2023 March 30]. Available from: https://doi.org/10.15406/oajtmr.2017.01.00006.
4. Tanwar YS, Kharbanda Y, Rastogi R, Singh R. Desmoplastic fibroma of bone: a case series and review of literature. Indian Journal of Surgical Oncology. 2018; 9(4): 585-591. [cited 2023 February 01]. Available from: https://doi.org/10.1007/s13193-018-0767-1.
5. Guruprasad Y, Chauhan S. Desmoplastic fibroma of the mandible. Medical Journal of Dr. D.Y. Patil University. 2015; 8(2): 254-257. [cited 2022 December 16]. Available from: https://www.mjdrdypu.org/text.asp?2015/8/2/254/153180.
6. Melzer RS, Lunelli DC, Patussi C, Sassi, LM. Surgical treatment of desmoplastic fibroma in the jaw using mandibular graft reconstruction – a case report. South Brazilian Dentistry Journal. 2018; 15(2): 130-134. [cited 2023 April 01]. Available from: https://www.researchgate.net/publication/330858979_Surgical_treatment_of_desmoplastic_fibroma_in_the_jaw_using_mandibular_graft_reconstruction_-_a_case_report/fulltext/5c58f489458515a4c7590f3a/Surgical-treatment-of-desmoplastic-fibroma-in-the-jaw-using-mandibular-graft-reconstruction-a-case-report.pdf.
7. Shekhar MG, Reddy RS, Ravikanth M, Lavanya R, Kumar M. Desmoplastic fibroma of the mandible: case report and review of literature. Primary Dental Journal. 2011; 18(3): 115-118. [cited 2023 March 12]. Available from: https://doi.org/10.1177%2F2050168411os1800305.
8. Nithya S, Sundaravel S, Divya U, Kameswara R. Desmoplastic fibroma – a rare case report. Journal of Oral and Maxillofacial Pathology. 2015; 19(2): 270. [cited 2023 March 19]. Available from: https://doi.org/10.4103/0973-029x.164564.
9. Summa A, Cerasti D, Crisi G, Ormitti F, Ventura E, Sabato M. Desmoplastic fibroma of the mandible: usefulness of CT and MRI imaging in Diagnosis and Treatment. 2010; 23(1): 109-113. [cited 2023 February 01]. Available from: https://doi.org/10.1177/197140091002300118.
10. Gersak MM, Lenghel LM, Baciut G, Moldovan I, Mitea H, Botar-Jid C, Vasilescu D, Botan E, Dudea SM. Ultrasonography of desmoplastic fibroma of the mandible; a case report. Medical Ultrasonography. 2015; 17(3): 407-410. [cited 2023 February 01]. Available from: http://dx.doi.org/10.11152/mu.2013.2066.173.mmg.
11. Evans S, Ramasamy A, Jeys L, Grimer R. Desmoplastic fibroma of bone: a rare bone tumor. Journal of Bone Oncology. 2014; 14(3): 77-79. [cited 2023 March 21]. Available from: https://doi.org/10.1016/j.jbo.2014.08.001.