ABSTRACT
Background: Fibrous dysplasia (FD) is a benign fibro-osseous lesion of the bone which can be monostotic FD and the polyostotic form. Polyostotic FD with café-au-lait spots of the skin and hormonal imbalances is called McCune–Albright syndrome. Besides, Mazabraud syndrome is characterized by polyostotic FD and intramuscular myxomas. FD has its onset during childhood or early adolescence and usually occurs within the first or second decade of life.

The mode of presentation of the FD of the jaw ranges from asymptomatic to dental anomalies, pain, and facial asymmetry. Given the clinical history and radiological assessment, cystic lesions have some important differential diagnoses which range from cystic ameloblastoma, fibrous dysplasia, aneurysmal bone cyst, and odontogenic keratoacyst.

Case report: This presents a 19-year-old female who presented to the maxillofacial surgeons’ clinic with a referral from a peripheral hospital. Her major complaint was right-sided facial swelling which was noticed 4 years before the presentation. A plain radiograph revealed an expansile lesion of the mid-right mandible appearing as a well-outlined, fairly oval multiloculated cystic radiolucent mass with multiple internal septations. A preliminary diagnosis of a complex cystic right jaw mass with benign features was made with possible differential diagnoses such as ameloblastoma, fibrous dysplasia, aneurysmal bone cyst, and odontogenic keratoacyst.

Finally, she had an excision biopsy with a histological diagnosis of fibrous dysplasia. The outcome of the surgery was satisfactory at one year review.

Keywords: Fibrous dysplasia (FD), Mandible, Magnetic resonance imaging (MRI)

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INTRODUCTION
Fibrous dysplasia (FD) is a congenital fibro-osseous disorder arising from sporadic mutation of the α-subunit of the G-stimulatory protein with resultant replacement, and distortion of bone by disorganized structurally weak fibrous tissue. The term 'fibrous dysplasia' was originally coined and described by Lichtenstein in 1938 as a disorder that is characterized by progressive replacement of normal bone by fibrous tissue. The pathogenesis of fibrous dysplasia is not fully understood. The available pieces of evidence point to post-zygotic mutation of the alpha (α) subunit of the G-stimulatory protein (GNAS) which leads to activation and overproduction of inappropriate cyclic adenosine monophosphate leading to the formation of new bone marrow stromal cells and associated impaired capacity to differentiate into mature osteoblasts, adipocytes, and haematopoietic cells. Yevgeniya et al reported that impaired haematopoietic cell production may manifest as haemorrhage and cystic changes which may be associated with a secondary aneurysmal bone cyst. The clinical manifestation of fibrous dysplasia of the jaw is that of a slow-growing painless mass that is hard in consistency. It may be associated with intermittent pain due to impingement on the neurovascular structure as it enlarges. The true incidence of fibrous dysplasia is not known but is estimated as 2.5 to 10% of all bone tumours. The disease manifests in different forms with differing nomenclatures: it can occur on a particular bone as monostotic FD—seen in 80% of the lesions, or multiple bones as polyostotic FD—which is also known as Jaffe-Lichtenstein disease for isolated fibrous dysplasia. McCune–Albright syndrome denotes the spectrum of fibrous dysplasia with associated extraskeletal abnormalities/endocrinopathies, while Mazabraud syndrome is used to describe fibrous dysplasia with associated intramuscular myxomas. Similarly, Cherubism, was equally considered a variant of fibrous dysplasia in the past which is now known to be a genetically distinct disease entity. Fibrous dysplasia commonly occurs at the long bones, craniofacial bones, ribs, and pelvic bones. Fibrous dysplasia of the mandible is rare and usually present as a monostotic form in 74%, as the polyostotic form in 13%, and as part of the craniofacial bone fibrous dysplasia in 13%. Cystic degeneration of fibrous dysplasia is an even rarer form of manifestation of the disease with a documented incidence of 10-25% of fibrous dysplasia. Thus, we present a case report and literature review of a large jaw mass in a 19-year-old female with features suggestive of benign cystic jaw mass on imaging and final histologic diagnosis of fibrous dysplasia. This report highlights the unusual location and imaging features of fibrous dysplasia.

CASE PRESENTATION: Miss D.J. is a 19-year-old female who presented to the maxillofacial surgeons’ clinic with a referral from a peripheral hospital. Her major complaint was right-sided facial swelling which she noticed 4 years before the presentation. She was normal until 4 years before presentation when she felt intermittent pain on her right cheek together with a small swelling on the same part of the jaw. Pain was mild initially but increased with the size of the swelling, intermittent, not radiating to any other part, and not relieved by analgesics. The swelling subsequently increased in size over the years. There was no ulceration, no loss of sensation, no prior history of trauma, and no history of tooth extraction. There were no abnormal nodules/swelling in any other part of her body, no history of drenching night sweats, or weight loss. No history of renal diseases, no history of chronic illness, and no area of abnormal pigmentation in her skin. She had a negative history of chronic cough or contact with an adult who had a medical history of chronic cough. The patient had no family history of similar mass or malignancy. She achieved menarche at 15 years old and has maintained a regular 28-day menstrual cycle. The patient had presented to a peripheral hospital where an attempt to excise the mass failed before she was referred for expert attention. She was worried due to the facial asymmetry and aesthetics. Examination revealed facial asymmetry with a hard non-tender mass on the right cheek as demonstrated in Figure 1 below. The mass has an irregular surface, no differential warmth, and no ulceration; not attached to the skin but inseparable from the underlying ipsilateral mandible. However, fine needle aspiration cytology (FNAC) was not done. The mass had an old healed surgical scarification (from a failed surgical attempt at the peripheral hospital). The submandibular, parotid, cervical lymph nodes and thyroid gland were not palpable, and no tenderness on these regions on palpation. The lips were complete and competent. Temporomandibular joints, palate, tongue, and buccal mucosa were clinically normal. The rest of the general and systemic clinical examination findings were also normal.
INVESTIGATIONS: The full blood count and urinalysis were normal. Retroviral status was negative. Serum electrolytes, creatinine, calcium, and alkaline phosphate levels were normal. A plain radiograph revealed an expansile lesion of the mid-right mandible appearing as a well outlined fairly oval multiloculated cystic radiolucent mass with multiple internal septations, cortical thinning, and a narrow zone of transition. There was associated endosteal scalloping, and mild soft tissue swelling around the affected part of the right mandible. There was no calcific density or tooth within the mass. The crown and root of the teeth in the adjacent alveolar ridge were spared. The left jaw and surrounding soft tissue shadows appear normal as demonstrated in Figure 2. Complementary ultrasonography showed cystic right jaw mass with hypoechoic fluid contents with hyperechoic strands and posterior acoustic enhancement, and eccentric irregular (jagged) focus suggestive of scalloped endosteal bony parts. There was no vascular flow on colour Doppler interrogation as demonstrated in Figures 3 and 4. There were no demonstrable parotid, submandibular, and cervical lymph node enlargements. The thyroid gland, the neck muscles, and vessels were sonographically normal. A preliminary diagnosis of a complex cystic right jaw mass with benign features was made. Cystic ameloblastoma, fibrous dysplasia, aneurysmal bone cyst, and odontogenic keratocyst were suggested as possible differential diagnoses. The patient underwent excisional tissue biopsy which was submitted for histopathological analysis. The specimen was received at the Department of Histopathology with 10% neutral buffered formalin and subsequently subjected to tissue processing which includes fixation, ascending grade of alcohol, clearing in xylene, and impregnation. After that, embedding, microtomy, and staining were done with haematoxylin and eosin. The slides were reviewed by a Consultant Anatomic Pathologist who finalized it as fibrous dysplasia. The histology was described as irregular woven bone arranged in Chinese letters and surrounded by hypocellular fibroblastic stroma with some myxoid areas; no mitotic figures and no cellular atypia were noted as shown in Figure 5. The patient was successfully operated upon with satisfactory post-operative outcome and regular on monthly follow-up for a year with no fresh complaints. ETHICAL ISSUES The informed consent of the patient for publication was obtained and the case report was conducted in compliance with the guidelines of the Helsinki Declaration on biomedical research in human subjects.
Figure 3: Gray-scale ultrasonographic image of right jaw showing iso-hypoechoic cystic mass with focus on expansile scalloped bony spicules like Chinese letter

Figure 4. Colour Doppler evaluation of the right jaw mass showing cystic lesion with a lack of peripheral vascular flow

Figure 5 x100Mag: The histologic section shows irregular trabeculae of woven bone merging to form complex shapes reminiscent of Chinese letters without osteoblastic rimming and surrounded by fibrovascular stroma.

DISCUSSION
Fibrous dysplasia of the jaw is considered a hamartomatous developmental anomaly of the jaw bone that is not periodontal in origin. Fibrous dysplasia is a congenital, metabolic, non-genetic disease of the bone comprising about 2.5% of the bone tumours and up to 7% of benign tumours of the facial and cranial bone with about 50% of Polyostotic fibrous dysplasia, and in 10-27% of monostotic fibrous dysplasia. Notably, among the craniofacial bones, the maxilla is more commonly involved than the mandible. The index case involved the mandibular bone.

Fibrous dysplasia is believed to be commoner among females and frequently reported in the younger age group with 75% occurring before 30 years and continues to regress after puberty. However, another study documented no gender predilection and that the lesion continues to advance till old age. This is in keeping with the index case whose lesion has been increasing progressively with no evidence of regression over the years despite being within the window period of regression after the age of puberty. The aetiology of fibrous dysplasia is unknown. It is believed to be a hamartomatous developmental abnormality characterized by irregularly distributed
spicules of bone in the cellular fibrous stroma with current plausible evidence implicating infectious or traumatic causes.\(^6,7,8\) The index case is supported by unknown aetiology as there was no clinical evidence of any causative agent and no history of trauma prior to the onset of the lesion. More importantly, this index patient had an expansile cystic jaw mass with multiple septation which is in keeping with the reports of Yevgeniya et al.\(^5\) The spectrum of imaging findings in fibrous dysplasia especially plain radiographs and computed tomography are classified into three broadly into: sclerotic, cystic, or mixed or ‘ground glass’ in appearance.\(^1,4\) It may appear as an expansile bony lesion with cortical thinning, and pathologic fracture. In addition, Computed tomography may demonstrate soft tissue involvement in Mazabraud syndrome which is mainly associated with polyostotic fibrous dysplasia, and show the extent of the lesion better than plain film radiography. Magnetic resonance imaging (MRI) also shows better soft tissue resolution and delineates the extent of the lesion better but does not show any specific findings. MRI shows variable \(T_1\) and \(T_2\) –weighted intensities and enhancement patterns depending on the amount of bony trabeculae, cellularity, collagen, cystic and haemorrhagic changes.\(^1,9\) The higher the amount of bony trabeculae, the lower the \(T_2\) signal intensity and vice-versa; fibrous dysplasia also contain cystic changes which appear brighter in \(T_2\)-weighted images.\(^1\) Fibrous dysplasia also shows a variable degree of enhancement on postcontrast \(T_1\)-weighted images: active lesions show avid enhancement while inactive lesions show mild enhancement. The patterns of enhancement include: patchy, central, rim, homogenous, or mixed.\(^1,9\)

Magnetic resonance imaging, therefore, does not help differentiate fibrous dysplasia from other confounding differential diagnoses. It is, however, helpful in assessing the complications of the lesions such as compression of neurovascular structures, malignant changes by use of diffusion-weighted imaging, and excluding secondary aneurysmal bone cyst.\(^5, 5, 10\) This index patient did not procure computed tomography image nor magnetic resonance imaging because the combined plain radiographic and ultrasonographic findings helped assess the bony and cystic nature of the lesion. Ultrasonographic evaluation of jaw masses is very helpful in establishing the cystic nature of fibro-osseous lesions, assessing the soft tissue components of the lesions, and detecting the cortical bony outlines-thickness, and vascularity of the lesions as documented by other reviewers.\(^3,12\) Sonographic findings in this index patient yielded important diagnostic information which included: iso-hypoechoic cystic mass with posterior acoustic enhancement, the focus of expansile scalloped cortical bony spicules with no significant vascular flow in colour. Doppler interrogation-suggestive of a benign lesion. These findings were in keeping with those documented by Shahidi et al and Gad et al.\(^3,12\)

The diagnosis of fibrous dysplasia of the jaw is established by histology. However, imaging evaluation is important in narrowing the differential diagnoses, ruling out or ruling-in possible complications like malignant transformation – which may occur in 2.5% of the cases.\(^1\) Imaging is also important for patient follow-up either in the pre- or post-surgical stage. More importantly, radiological documentation of cystic changes in histologically diagnosed fibrous dysplasia has been documented by other reviewers which is consistent with the index patient.\(^13,14\) The index patient underwent surgical excision with a histological diagnosis of fibrous dysplasia. She is being managed conservatively and on monthly follow-up for a year with satisfactory outcomes.

CONCLUSION

The mandible is a rare location of fibrous dysplasia generally but a common location of craniofacial fibrous dysplasia. Imaging findings and clinical history are suggestive of the lesion and will help to direct the radiologists’ attention. It is therefore important that radiologists include fibrous dysplasia in the list of considerations in differential diagnoses of posterior lower masses. More importantly, the specimen must be subjected to histological examination which is the goal standard of tissue diagnosis.

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Conflict of interest
None declared

REFERENCES