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(CASE REPORT)

Craniofacial fibrous dysplasia mimicking sinunasal mucocele: Case report

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Abstract

Background: Fibrous Dysplasia is a congenital, non-inherited, benign intra-medullary bone lesion in which the normal bone marrow is replaced by abnormal fibro-osseous tissue. It can manifest in axial and extra -axial skeleton and tissues. The wide range of clinical manifestations from skeletal changes to soft tissues swellings and skin discoloration makes it a disease of note in the clinical settings. Some of its complications include malignant transformation, gastrointestinal reflux to cardiac involvement with arrhythmias and even sudden death. Until recently with advent of modern imaging modalities, it was a disease of diagnostic debacle as it was often misdiagnosed .This is a case report of a 17 year old male who was misdiagnosed as sinunasal mucocele until histology proved it to be fibrous dysplasia. **Objective:** The objective of the report is to draw attention to the diagnostic challenge posed at CT imaging.

Methods: This case report was carried out at University College Hospital Ibadan.

Results: Showed right sided facial swelling measuring about 15x10cm. The mass was firm to hard in consistency, non-tender and not attached to the overlying skin. There was no loss of sensation in the skin over it. The right eye was displaced superiorly and laterally. A nasal aspect of the mass was seen to occupy the whole of the ipsilateral nasal cavity which it occluded deviating the nasal septum to the left and narrowing the left nasal cavity. It was fleshy and showed contact bleeding. The attending ENT Surgeons made a working diagnosis of right sinunasal tumour with possible malignant transformation. The radiographs of the sinuses showed expansile sclerotic lesion involving the nasal bones, the right maxillary bone, right ethmoidal and maxillary sinuses. The inferior aspect of the right frontal sinus was also affected. A cranial computed tomography (CT) scan (Fig 2-4) showed a multi-loculated hypodense mass with mildly enhancing septations (HU16) within the right frontal, ethmoidal, sphenoidal and maxillary sinuses. There was associated expansion of the affected sinuses with thinning of their cortices. No evidence of aggressive bony destruction was seen. The radiologists made a diagnosis of benign sinunasal tumour most likely mucocele. After right maxillectomy, histology proved the lesion to be fibrous dysplasia. This report goes further to support the need for application of more imaging modalities especially the modern and higher slices CT scanner, higher Tesla(2T)MRI , PET and fusion CT/PET scanners in arriving at radiological diagnosis that will be more compatible with the pathological condition of the patients as against what was in place at the time of this case report .Radiologists are also encouraged to have a high index of suspicion for the disease and to explore the possibilities of the differentials in order not to miss the diagnosis of FD

Keywords: Computed Tomography (CT); Magnetic Resonance Imaging (MRI); Fibrous Dysplasia; Mucocoele

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1 Introduction

Fibrous dysplasia (FD) is a rare condition representing 2.5-5% of benign bone lesions[1] .FD was first described in 1942 by Lichtenstein and Jaffe [2] ; therefore it is sometimes referred to as Lichtenstein-Jaffe disease.It is a developmental anomaly of the bone-forming mesenchyme that manifests as a defect in osteoblastic differentiation and maturation resulting in poorly organized, structurally unsound fibrous tissue by a process of metaplasia [3.] It results from sporadic mutation of the alpha subunit of the Gs stimulatory protein[3]. The disorder can be monostotic (involving a single bone) or polyostotic (involving multiple bones) [2].It can result in pain, deformity, fractures, or abnormalities in bone mechanics[4]. When FD occurs in the setting of other extraskeletal abnormalities, skin pigmentation, and endocrine dysfunction, the resulting syndrome is named McCune-Albright syndrome[5].

Endocrinopathies such as hyperthyroidism, hyperparathyroidism, acromegaly, diabetes mellitus, and Cushing syndrome have been reported in association with FD[2]. When FD occurs -and this is rare, in association with solitary or multiple soft-tissue myxomas, the condition is called Mazabraud syndrome[6].

FD is known to have a higher incidence in West Africa than the whole of Europe and it may affect any bone[7].The purpose of this case report is to highlight the diagnostic dilemma posed by this lesion on computed tomographic study (CT).

2 Case report

O. O. is a 17 year-old male Senior Secondary School student who presented at the Ear, Nose and Throat (ENT) Clinic for right facial swelling and nasal obstruction for 8 years. The swelling was painless and slowly growing. There was associated purulent right eye discharge but vision was not impaired. At the same period, patient started having nasal congestion that progressed to obstruction and eventually involved both sides associated with anosmia. There was recurrent provoked epistaxis which was scanty. There was no gum swelling, bleeding or teeth loosening but right –sided frontal headache which started 21 weeks before presentation. Patient was referred to University College Hospital (UCH) Ibadan from Ogun State University Teaching Hospital.

Examination showed a right sided facial swelling measuring about 15x10cm. The mass was firm to hard in consistency, non-tender and not attached to the overlying skin. There was no loss of sensation in the skin over it. The right eye was displaced superiorly and laterally. A nasal aspect of the mass was seen to occupy the whole of the ipsi lateral nasal cavity which it occluded deviating the nasal septum to the left and narrowing the left nasal cavity. It was fleshy and showed contact bleeding. The dentition was however intact. There were no skin pigmentations, swellings in other parts of the body. The remaining systems were essentially normal. The endocrine profile of the patient was normal.

A working diagnosis of a sinunasal tumour was made. The differential diagnosis was fibro-osseous disease (fibrous dysplasia) with a likely malignant transformation. The radiographs of the sinuses showed expansile sclerotic lesion involving the nasal bones, the right maxillary bone, right ethmoidal and maxillary sinuses. The inferior aspect of the right frontal sinus was also affected. A cranial computed tomography (CT) scan (Fig 2-4) showed a multi-loculated hypodense mass (HU16) within the right frontal, ethmoidal, sphenoidal and maxillary sinuses. There was associated expansion of the affected sinuses with thinning of their cortices. No evidence of aggressive bony destruction was seen. The mass in the nasal cavity crossed the midline with complete obliteration of the ipsilateral nasal cavity and narrowing of the contralateral nasal cavity. The left maxillary sinus showed complete opacification with lateral deviation of its medial wall. There was associated anterolateral displacement of the right orbit. The contrast slices showed some enhancement of the internal septae within the mass (Fig 3). There was a slight angulation and a small defect in the bony floor of the anterior cranial fossa on the right (Fig 4). However, no intracranial extension of the mass was seen and the visualized brain parenchyma appeared within normal limits. The radiological features were highly suggestive of a long standing benign lesion, most likely an extensive mucocele. At surgery the mass was located mainly in the maxilla and a right total maxillectomy was done. Histology result came out with diagnosis of fibrous dysplasia. Patient did well post surgery and was a subsequently discharged to continue follow up in the surgical clinic at UCH Ibadan.

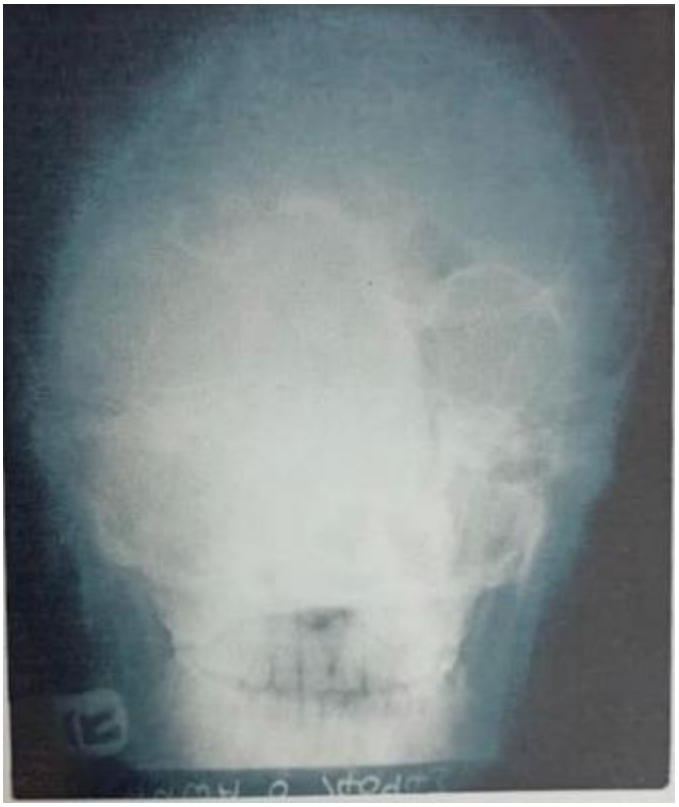

Figure 1 Plain radiograph (occipitofrontal view) showing expansile sclerotic lesion in the right facial region displacing the medial wall of right orbit laterally

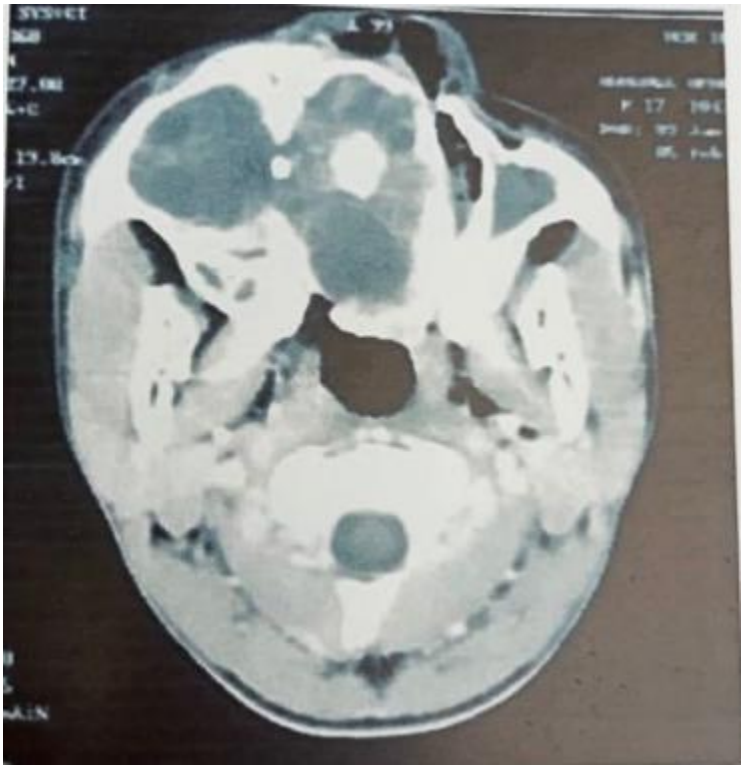

Figure 2 A contrast enhanced axial slice of the cranial base showing hypodense lesion in the right maxillary sinus and right nasal cavity

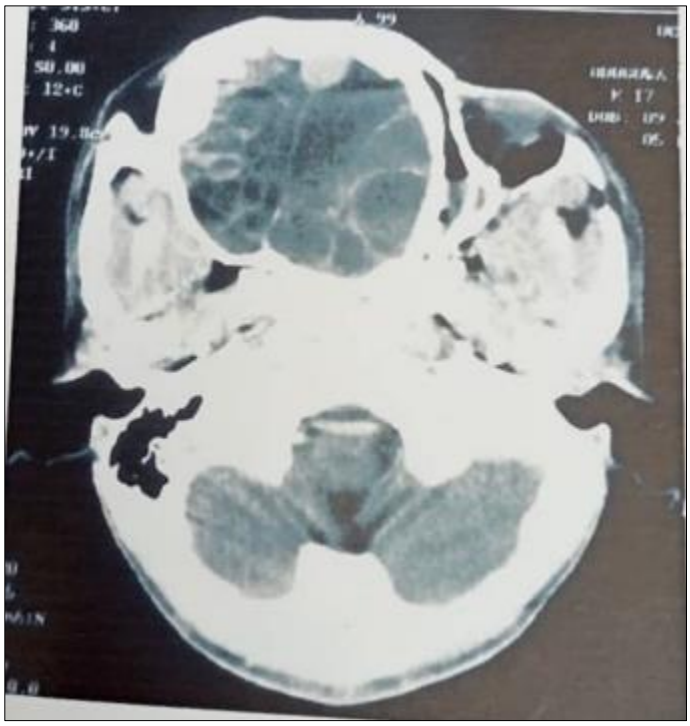

Figure 3 A contrast enhanced axial slice showing hypodense lesion with multiple partial septations in an expanded right ethmoidal sinus with sclerotic walls. The left ethmoidal sinus is compressed

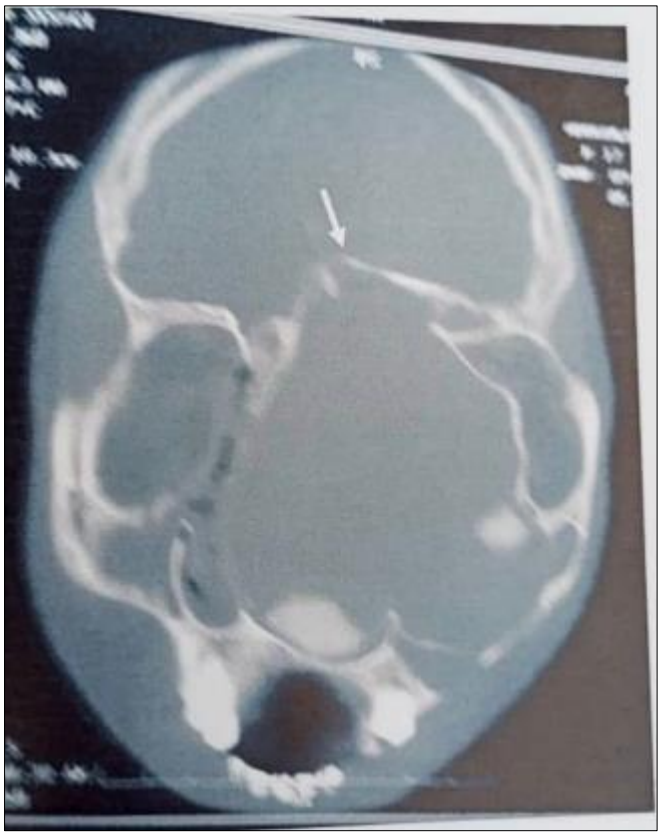

Figure 4 A Coronal image (bone window) showing elevation and angulation of the cribiform plate with a small breach (↓)

3 Discussion

FD affects males and females equally, except in McCune-Albright syndrome where females are more frequently affected [8].The age of skeletal onset of FD is between 10 and 30 years [2]. The lesion resolves after skeletal maturity but may be seen in old age[2] . Our patient is 17 year old male. His age falls within the 1st three decades widely noted in the literature [7,9,10.]

Our case shows only skeletal manifestations: in the cranial and facial bones. There were no skin pigmentations, swellings of the soft tissues or endocrine abnormalities. It is therefore considered a polyostotic variety of FD as against monostotic, where only one bone is involved which are known to likely enlarge in adult life [9,11]. Also, there was no association with hyper pigmentation of the skin, early skeletal maturation or precocious puberty hence no qualification for consideration as McCune-Albright disease. The finding of FD involving the craniofacial bone in our case report agrees with earlier report by Odeku et al [12] and Obisesan et al [7] that craniofacial and jaw FD appeared to be commoner in Nigerians than the long bone variety . This however contradicts report by Wei & Co[13] who found the long bone variety to be commoner. This may not be unconnected with similar reason why lymphoma of the jaws(Burkitt) is commoner in Nigerians than the Caucasians. Apart from heredity; dietary and environmental factors may be contributory. In this case, there was involvement of the maxillary bone, the sphenoid bone, the turbinate, and the cribiform plate on the right side but the mandible was spared. Less involvement of the mandible was similarly observed in different studies by Daramola & Co, [14] Odeku[12] and Obisesan[7] in the same environment where they also found the maxilla to be involved in craniofacial FD more than the mandible as similarly reported by Singh & co[15].

The plain radiographs (paranasal sinuses views) of our case showed expansile sclerotic lesion involving the nasal bones, the right maxillary bone, right ethmoidal and maxillary sinuses. The inferior aspect of the right frontal sinus was also affected. This is similar with the diffuse sclerosis of FD in the literature[2,7.] Diffuse sclerosis is the 3rd commonest radiographic type of craniofacial FD after peau d orange and whorled plaque-like types.[2,7] The bone lesions can be classified as cystic, sclerotic, or mixed. A typical FD lesion in the axial skeleton appears as radiolucent ground-glass matrix that is usually smooth and homogeneous. The FD lesions usually cause cortical thinning because of enlarged fibro-osseous masses. [2]

The CT finding of hypodense lesion with HU of 16 with enhancing septation suggests a rather cystic nature contradicting the finding of diffuse sclerosis seen in the plain radiograph. This could be attributable to underexposure of the film and the poor resolution due to superimposition of structures characteristic of plain radiograph. The non correlation of the plain radiographic and CT findings can again be attributed to the improved resolution of CT images. This improvement has been magnified in more modern CT scanners as against the earlier version that was in use when these CT images were obtained for the patient in UCH Ibadan in early 2000.

The absence of periosteal reaction and soft tissue mass in this case report agrees with the finding in the literature [6,7.] The contrary finding is the angulation and defect in the cribiform plate suggesting fracture. This fracture could be from pressure effect. The risk of fracture even in the face of limited disease in the bone affected by fibrous dysplasia has been reported, even though the actual fractures documented were noted in the long bones of the limbs[2].

Although in the present case, there was an extension of the lesion into the orbit displacing the eye ball anterolaterally, the vision was not impaired. This contradicts a report by Sadigh & Co [16] who found in a 27 year-old man with FD lesions involving the left frontal bone, left side of the sphenoid, left posterior ethmoid and ethmoidal sinuses, and circumferential involvement of the orbit(except for its inferior wall) with compressive optic neuropathy in the left eye. The eye had the best corrected vision of 6/30 which turned to 6/6 after endoscopic transnasal orbital and optic canal decompression. Cranial nerve palsies are known complications of fibrous dysplasia[9].

The intringuing thing about our case was that mucocele could give features of opacification and expansion of affected paranasal sinuses which in plain radiograph may look similar to FD.

The CT findings of mucocele in literature shows that affected sinus is completely opacified and the margins are expanded and usually thinned. Areas of complete bone resorption may be present, resulting in bony defect and extension of the 'mass' into adjacent tissues [17,18]. Peripheral calcification is sometimes seen [17]. Following administration of contrast, only peripheral enhancement (if any) is seen. Van tassel, Lee, Jing, et-al (1989, as cited in Tran, 2023) posited that the content of the sinus is variable, depending on the degree of hydration, ranging from near-water attenuation to hyper-attenuation as secretions become increasingly thick and desiccated [18].These findings agree with ours except that in our case, there were no calcifications; and there were septations giving cystic appearance rather than the non-septate appearance in mucocele. Also, there was inclusion of FD in the differentials of the working diagnosis from the requesting Physicians

though it appeared attempts were not made adequately to exclude the FD as stated in the differentials which could have swayed the radiologic diagnosis in favour of FD instead of the mucocele. However, with more imaging techniques-higher slices CT scanner and MRI this diagnostic dilemma could have been easily resolved. Higher slices CT scanner and MRI were not available in the centre at the time of the case report.

Sarcomatous change is also known to be a complication of FD with prevalence of 0.4-4% [2]. Records show that the rate of malignant transformation is higher for polyostotic than for monostotic lesions; it is commoner in craniofacial region, followed by femur, tibia, and pelvis; and especially in patients with previous irradiation [19,20]. Our case shows no evidence of Sarcomatous change in the radiological and histological report. It is of note that increased risks of breast and prostate cancers have also been observed in people with FD in the literature[21].This should suggest the need for long term follow up of patients with FD.

4 Conclusion

Until recently with the advent of modern imaging modalities, FD was a disease of diagnostic debacle as it was often misdiagnosed. This is seen in this case of a 17 year-old male student with eight years history of right facial and intranasal mass presented. Though the clinical and radiological features were more in favour of paranasal sinus mucocele, however, the histology revealed fibrous dysplasia. There is need for high index of suspicion and for Radiologists to explore the differentials of the disease in question with a view to excluding them in order to arrive at a more accurate diagnosis. The use of multiple imaging modalities especially the more modern ones with higher diagnostic capabilities cannot be over emphasized. Multidisciplinary approach is also recommended for best quality patient-centred service delivery. These will go a long way in saving the time and cost of treatment to the patient, care givers as well as the community at large.

Compliance with ethical standards

Acknowledgement

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Disclosure of conflict of interest

No conflict of interest to be disclosed.

Statement of ethical approval

The study involves use of human subject, but being a case report, obtaining ethical clearance was not applicable.

Statement of informed consent

Informed consent was obtained from participants included in the study.

References

[1] Robinson C, Collins MT, Boyce AM. (2016).Fibrous dysplasia/McCune-Albright syndrome: clinical and translational perspectives. Curr Osteoporos Rep. 14(5):178-86.

[2] Gitto,L, Zaccarini, DJ. (2021).Fibrous Dysplasia Pathology, https://emedicine.medscape.com/article/1998464-overview#a2

[3] Anand, MKN.2020. FibrousDysplasi-Imaging. https://reference.medscape.com/article/389714-overview#a1

[4] Majoor BCJ, Traunmueller E, Maurer-Ertl W, Appelman-Dijkstra NM, Fink A, Liegl B, Hamdy NAT, Dijkstra PDS, Leithner A. (2019). Pain in fibrous dysplasia: relationship with anatomical and clinical features. Acta Orthop. 90(4):401-5.

[5] Kushchayeva YS, Kushchayev SV, Glushko TY, Tella SH, Teytelboym OM,Collins MT,and Boyce AM, (2018). Fibrous dysplasia for radiologists: beyond ground glass bone matrix. Insights Imaging. 9 (6):1035-1056.

[6] Majoor BCJ, van de Sande MAJ, Appelman-Dijkstra NM, Leithner A, Jutte PC, Vélez R, Perlaky T, Staals TEL, Bovée JVMG, Hamdy NAT, Dijkstra SPD.(2019). Prevalence and clinical features of Mazabraud syndrome: a multicenter European study. J Bone Joint Surg Am. 101(2):160-8.

[7] Obisesan A.A., Lagundoye S.B., Daramola J.O., Ajagbe H.A. Oluwasanmi J. O.(1977). The Radiologic features of fibrous dysplasia of the craniofacial bones. Oral surg. 44:949-959.

[8] Corica D, Aversa T, Pepe G, De Luca F, Wasniewska M.(2018) Peculiarities of precocious puberty in boys and girls with McCune-Albright syndrome. Front Endocrinol (Lausanne). 9:337.

[9] Sweeney K, Kaban LB.(2020). Natural History and Progression of Craniofacial Fibrous Dysplasia: A Retrospective Evaluation of 114 Patients From Massachusetts General Hospital. J Oral Maxillofac Surg.May 30. https://read.qxmd.com/read/32619461/natural-history-and-progression-of-craniofacial-fibrous-dysplasia-a-retrospective-evaluation-of-114-patients-from-massachusetts-general-hospital?redirected=slug

[10] Alkhaibary A, Alassiri AH, Alsalman M, Edrees S. (2019). Unusual presentation of fibrous dysplasia in an elderly patient. J Radiol Case Rep. 13(2):26-33.

[11] Couturier A, Aumaître O, Gilain L, Jean B, Mom T, André M.(2017). Craniofacial fibrous dysplasia: A 10-case series. Eur Ann Otorhinolaryngol Head Neck Dis. 134 (4):229-235.

[12] Odeku E., Martinson D.F., Akinosi J.O.(1969). Craniofacial fibrous dysplasia in Nigerian Africans. Int. Surg 7; 96-103.

[13] Wei WJ, Sun ZK, Shen CT, Zhang XY, Tang J, Song TH , Qiu ZL, Luo QY (2017).Value of 99mTc-MDP SPECT/CT and 18F-FDG PET/CT scanning in the evaluation of malignantly transformed fibrous dysplasia. Am J Nucl Med Mol Imaging. 7(3):92-104.

[14] Daramola J.O., Obisesan A.A., ; Lagundoye S.B., Oluwasanmi J.O.(1976). Fibrous dysplasia of the jaws in Nigeria. Oral Surg. 42:290-300.

[15] Singh V, Gupta K, Salunke P. (2019). Monostotic craniofacial fibrous dysplasia: report of two cases with interesting histology. Autops Case Rep. 9(2):e2018092.

[16] Sadigh, SL, Ozer,S, Bulut,EG, and Yavas,GF (2022).Fibrous dysplasia: A rare cause of optic neuropathy. Taiwan J Opthalmol 12(3):364-369.

[17] Mafee MF, Valvassori GE, Becker M. (2004)Imaging of the head and neck. George Thieme Verlag. ISBN:1588900096. Read it at Google Books - Find it at Amazon

[18] Tran, M. (2023) Paranasal sinus mucocele. https://radiopaedia.org/articles/paranasal-sinus-mucocele-1?lang=us#nav_radiographic-features

[19] Kinnunen AR, Sironen R, Sipola P.(2020). Magnetic resonance imaging characteristics in patients with histopathologically proven fibrous dysplasia-a systematic review. Skeletal Radiol. 49(6):837-45.

[20] Li Z, Raynald, Wang Z, Qian H.(2020). Malignant transformation of craniofacial fibrous dysplasia: a systematic review of overall survival. Neurosurg Rev. 43(3):911-21.

[21] Javaid MK, Boyce A, Appelman-Dijkstra N, Ong J, Defabianis P, Offiah A, Arundel P, Shaw N, Pos VD, Underhil A, Portero D, Heral L, Heegaard AM, Masi L, Monsell F, Stanton R, Dijkstra PDS, Brandi ML, Chapurlat R, Hamdy NAT, Collins MT.(2019)Best practice management guidelines for fibrous dysplasia/McCune-Albright syndrome: a consensus statement from the FD/MAS international consortium. Orphanet J Rare Dis. 14(1):139.