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FAMILIAL FORM OF CRANIOFACIAL FIBROUS DYSPLASIA: A CASE REPORT .

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ABSTRACT

Fibrous dysplasia is a benign disease of bone that may affect single or multiple bones. Craniofacial fibrous dysplasia may occur in a familial manner. This report deals with a patient who presented with multiple bony craniofacial prominences; with a positive family history of similar lesions. The lesions started to appear at his early teenage years and new lesions continued to arise and grow till 18 years of age. The lesions were expansile causing esthetic impairment. The case was diagnosed as a familial form of craniofacial fibrous dysplasia. Surgical recontouring was performed to the lesion that formed the patient's chief complaint. Later on the fibro-osseous lesion crept into the pulp canals of the related teeth causing pulpitis. Craniofacial fibrous dysplasia presentation varies according to the area affected, but it generally has a favorable diagnosis. However, we do recommend that periodic vitality testing be performed for teeth in proximity of the lesion.

Keywords: Craniofacial fibrous dysplasia, Endodontic, Esthetics, Facial asymmetry, GNAS1 protein, Pulpitis.

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ABSTRACT

Fibrous dysplasia is a benign disease of bone that may affect single or multiple bones. Craniofacial fibrous dysplasia may occur in a familial manner. This report deals with a patient who presented with multiple bony craniofacial prominences; with a positive family history of similar lesions. The lesions started to appear at his early teenage years and new lesions continued to arise and grow till 18 years of age. The lesions were expansile causing esthetic impairment. The case was diagnosed as a familial form of craniofacial fibrous dysplasia. Surgical recontouring was performed to the lesion that formed the patient's chief complaint. Later on the fibro-osseous lesion crept into the pulp canals of the related teeth causing pulpitis. Craniofacial fibrous dysplasia presentation varies according to the area affected, but it generally has a favorable diagnosis. However, we do recommend that periodic vitality testing be performed for teeth in proximity of the lesion.

Keywords: Craniofacial fibrous dysplasia, Endodontic, Esthetics, Facial asymmetry, GNAS1 protein, Pulpitis.

INTRODUCTION

Fibrous dysplasia (FD) is a bone disease in which normal bone is replaced by bony and fibrous tissue.[1] Fibrous dysplasia has been proven to be caused by a postzygotic somatic mutation of the *GNAS-1* (Gunaine Nucleotide binding protein alpha stimulating activity polypeptide 1) gene.[2] However, Familial form of fibrous dysplasia has been rarely reported. In these familial cases, mutation of GNAS-1 (of fibrous dysplasia) and of 4p16 (of Cherubism) were not detected.[3] We report here a teenage patient who was diagnosed as a familial form of craniofacial fibrous dysplasia, causing mainly esthetic impairment. Craniofacial fibrous dysplasia presentation varies according to the area affected, but it generally has a favorable

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diagnosis. He underwent surgical recontouring but later developed further fibro-osseous lesion in the pulp canals of the related teeth causing pulpitis. Hence, we recommend that periodic vitality testing be performed for teeth in proximity of the lesion.

CASE REPORT

An 18-year-old male patient presented with a chief complaint of a bony swelling in the lower left posterior area affecting his facial symmetry. He reported that it had increased in size over a 5 months period. He was only bothered with its disfiguring nature but had no other complaints such as facial numbness or pain.

The patient gave a history of having had a similar maxillary lesion at the age of 12, which was excised, but the histopathology analysis results were lost. He also reported the growth of multiple bony swellings in his skull at the same period of time. These bony swellings had a progressive nature, till they reached a certain size, at which point they became static.

Family history revealed the patient's father and grandfather to have had similar multiple bony swellings in the skull. Otherwise, there was no significant medical, family or personal history.

Extraoral examination revealed the presence of 4 bony hard swellings in the calvarium. While intraoral examination showed a solitary oval hard swelling in the left mandibular alveolar bone, apical to the lower left premolars and the first molar, expanding the buccal plate of bone. The swelling size was 3cm (antero-posteriorly) x 2 cm (corono-apically). The localized swelling was non-tender and its covering mucosa did not show any abnormality.

Computed tomography (CT) imaging of the skull done at 12-years of age showed a well-defined bony lesion at the right maxillary bone with spotty ground glass calcifications. The expansile lesion encroached on the naso-pharynx posteriorly; the right nasal fossa medially deviating the nasal septum; elevated the floor of the right orbit superiorly and depressed the right aspect of the hard palate inferiorly. Four other expansile bony swellings in the calvarium were shown in the CT (Figure 1a). The CT also detected other lesions at the left ethmoid sinus, left maxillary bone and greater wings of sphenoid. Scanning of the whole skeleton for further bony lesions had been performed using radiographs of the axial skeleton followed by MDP bone scanning; revealing the only activity to be in the calvarium and the maxillary bone.

Cone-beam CT of the mandible showed an expansile bony radiolucent lesion with wispy radiopacities traversing through it. The lesion extended from the mesial surface of the lower left canine to the distal root of the lower left second molar. The expansile lesion did not cause root resorption or displacement. However, it displaced the inferior alveolar canal in an apical direction, expanded the buccal plate of bone periapical to the lower left first molar and perforated the lingual plate of bone. (Figure 1b to f)

Surgical recontouring was performed for esthetic reasons in accordance with the ethical standards of the institution and was done after obtaining the patient's informed consent. No further intervention was needed due to absence of any functional impairment (Figure 2). Histopathology of the shaved lesion showed interlacing bony trabeculae merging with the normal buccal plate of bone with focal osteoblastic rimming separated by fibrous stroma (Figure 3) favoring a diagnosis of fibrous dysplasia (FD).

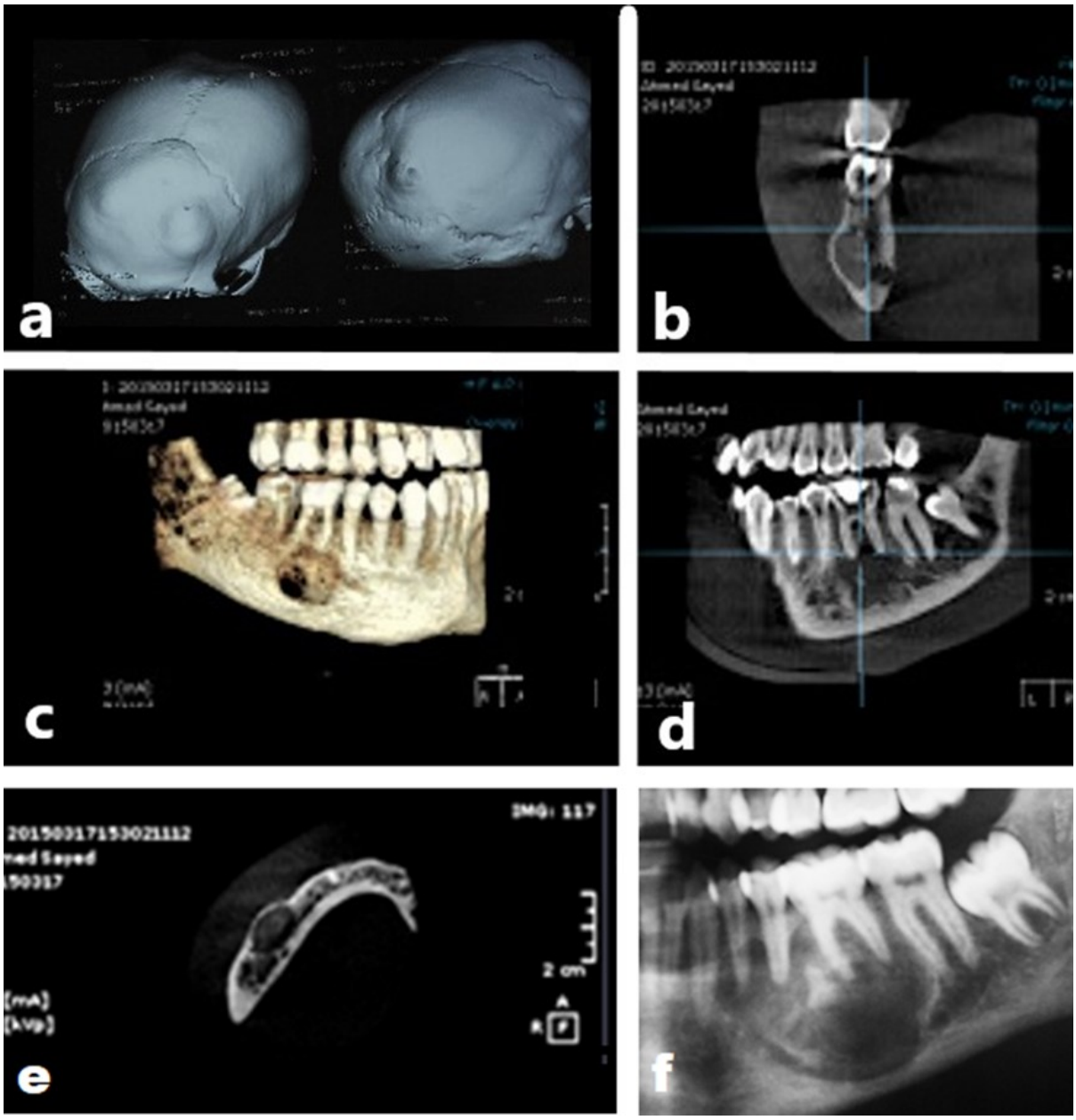

Figure 1: (a) A CT scan of the skull showing bony swellings in the frontal bones, the right parietal bone, the pterion and near the lambdoid suture; (b), (c) (d) and (e) CBCT cuts showing the lesion related to the lower left first molar and extending apically toward the lower border of the mandible; (f) Panoramic radiograph showing the lower left premolar-molar area where a radiolucent area with interrupted cortication and a radiopaque fragment inside.

At post-operative outpatient follow-up, the patient reported satisfactory esthetics with no signs of recurrence. The only complaint that arose one year later was pain related to the lower left first molar indicating endodontic treatment. However, the canals were obliterated by fibro-osseous tissue from the periapical bony lesion; indicating tooth extraction. Prognosis is favorable as FD lesions normally cease growth after puberty. However, the patient was advised of the need for long-term follow-up in case of malignant transformation.

DISCUSSION

Fibrous dysplasia is a benign condition where normal bone is replaced by fibrous tissue that can present as monostotic or polyostotic.[1] Craniofacial FD is a variant of the polyostotic

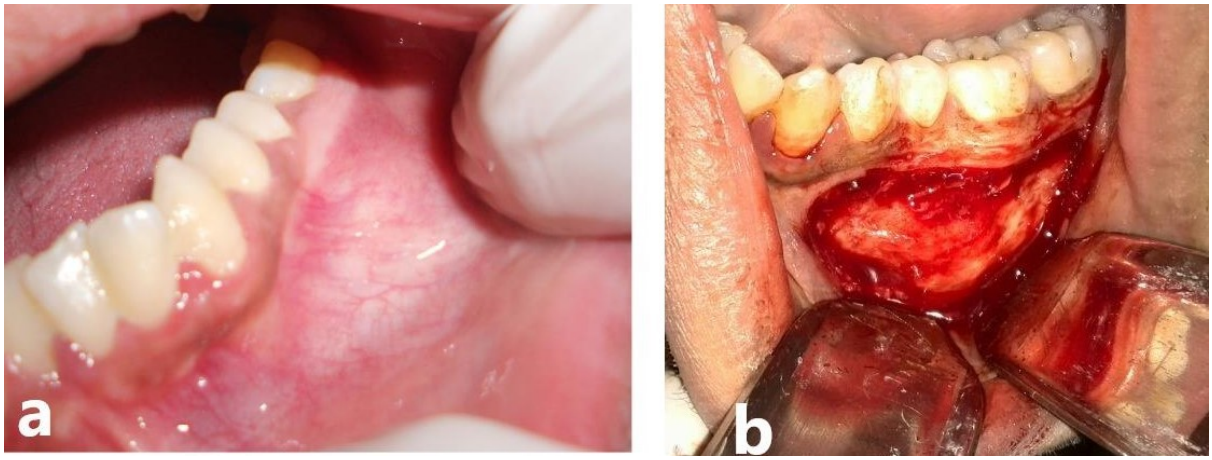

Figure 2: (a) preoperative clinical picture showing fusiform bony swelling in the left mandibular area with normal covering mucosa obliterating the buccal vestibule; (b) intraoperative picture showing the bony swelling after flap elevation and before surgical recontouring.

type. FD is a non-inherited disorder caused by a postzygotic mutation of the GNAS gene.[1] Interestingly, the patient's father and grandfather had similar lesion, suggesting a familial pattern or possible inherited susceptibility of acquiring the causative mutation. FD can appear as a deformity or can displace adjacent structures such eyes and teeth.[2] In this instance, the fibroosseous tissues caused an expansion of the buccal plate of bone and subsequent facial asymmetry. The aberrant tissues are generally invasive in nature and extended into the root canal of the lower left first molar obliterating the canal space.

Surgical treatment depends on many factors such as the involved site, rate of growth, patient's general health and preference.[4] Lesions affecting cranial base, mandible and maxilla are treated by conservative excision or shaving.[5] In the presented case, the expansion indicated that the disease while not aggressive was still active, which ideally should be left alone until skeletal maturity has been reached. However, the patient desperately demanded the surgery in order to pass a fitness test for college. Thus, surgical recontouring was done after informing the patient of the possibility of recurrence of the growth. A year later, disease activity flared up on the recontoured side. Fortunately, fibro-osseous proliferation was redirected up the root canals of the lower left first molar with no recurrence of facial asymmetry. By the

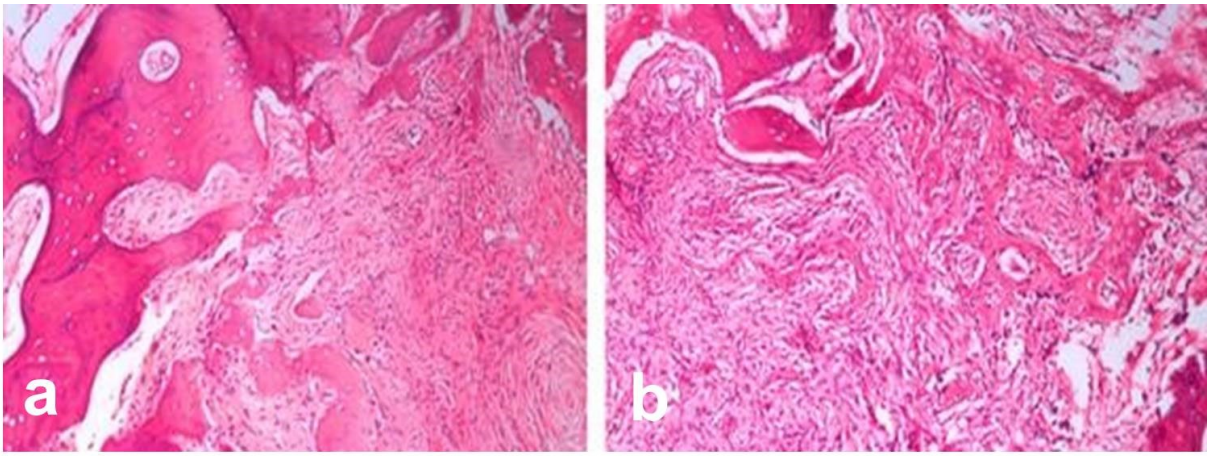

Figure 3 (a) and (b) Hematoxylin-eosin stained histopathological picture showing bony trabeculae with focal osteoblastic rimming separated by fibrous stroma, irregular woven bone. Bony components show areas of bone maturity, reversal lines and areas of new bone formation. Fibrous component shows areas of activity, fibrous swirling and bundling. (x100)

time the patient was complaining of pain indicating irreversible pulpitis, the pulp canals were half obliterated by the fibro-osseous tissues causing endodontic treatment to be impossible.

CONCLUSION

In cases where fibro-osseous lesions occupy the periapical vicinity of teeth, we recommend periodic vitality testing of these teeth with a frequency directly proportional to the rate of the lesional growth. This will be crucial to save the involved teeth as later on conservative treatment may be inapplicable.

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CONFLICT OF INTEREST

The authors declare no conflict of interest.

ETHICAL APPROVAL/INFORMED CONSENT

The treatment performed in the presented care is the standard of care and is in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1964 and later versions. For the application of treatment and publication of the case report, the patient signed an informed consent.

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