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CASE REPORT

Craniofacial fibrous dysplasia: A case report and literature review

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ABSTRACT

Fibrous dysplasia (FD) is a rare osseous pathology of unknown origin in which normal bone is replaced by fibro-osseous tissue. Recent research has linked FD to a somatic mutation in the protein transcription of the GNAS1 gene, which leads to an increase in intracellular cyclic adenosine monophosphate. FD represents 3% of all bony tumors and over 7% of all non-malignant bone tumors. FD has various clinical presentation groups such as the monostotic, craniofacial and polyostotic forms, and the McCune-Albright syndrome. We present a craniofacial FD case of a 45-year-old female patient, who underwent surgical treatment many times.

Key Words: CT, Fibrous, Dysplasia, Monostotic, Polyostotic, Craniofacial, MRI

1. INTRODUCTION

Fibrous Dysplasia (FD) is a relatively rare osseous disease of unknown etiology, where the normal bone is changed by fibrous tissue, bony metaplasia and newly formed poorly calcified bone. The majority of FD cases are found between 10-30 years old, has the same prevalence rate in males and females and shows variable radiographic features in relation with the level of maturation, which determines of the imaging characteristics. FD can be monostotic or, more rarely, polyostotic. Affection of the head osseous architecture, result in an evident cosmetic abnormality. The treatment of craniofacial FD should be focused on the relief of aesthetics and functional problems.

2. CASE REPORT

We present a craniofacial FD case of a 45-year-old female patient, without family medical history, who underwent sur-

gical treatment from 15 years ago. The periodical CT scan control, in bone window, revealed a craniofacial expansive bones lesions with heterogeneous density. The right ethmoid and maxillary sinuses had been totally occupied (see Figures 1-2). Additionally curved reformation of CT and 3D osseous volume rendering were created (see Figure 3). Clinically, a great facial deformity was observed, with some neurological symptoms like atypical facial pain and local lumps.

In addition, MRI was performed, showing an expansive process, well delimited, T1-weighted MRI showed a mildly hypointense, solid mass centered in the right craniofacial region, not observed intracranial invasion. T2-weighted MRI showed a mildly hyperintense mass, invading the right orbit with anterior and lateral displacement of the globe. Heterogeneous avid enhancement of the mass was seen in the post contrast fat-saturated images (see Figure 4).

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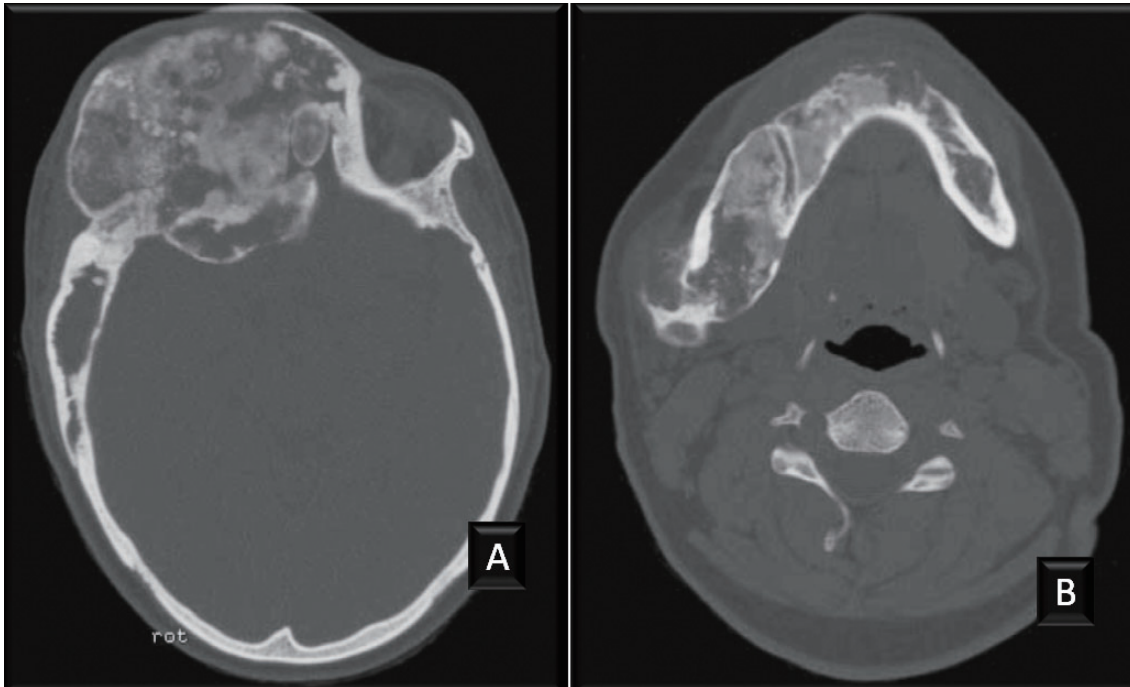


Figure 1. CT scan, bone window and axial view, showing (A) involvement of the frontal bone and (B) mandible. One can see a thin and sometimes perforated cortex and irregular and ill-demarcated borders.

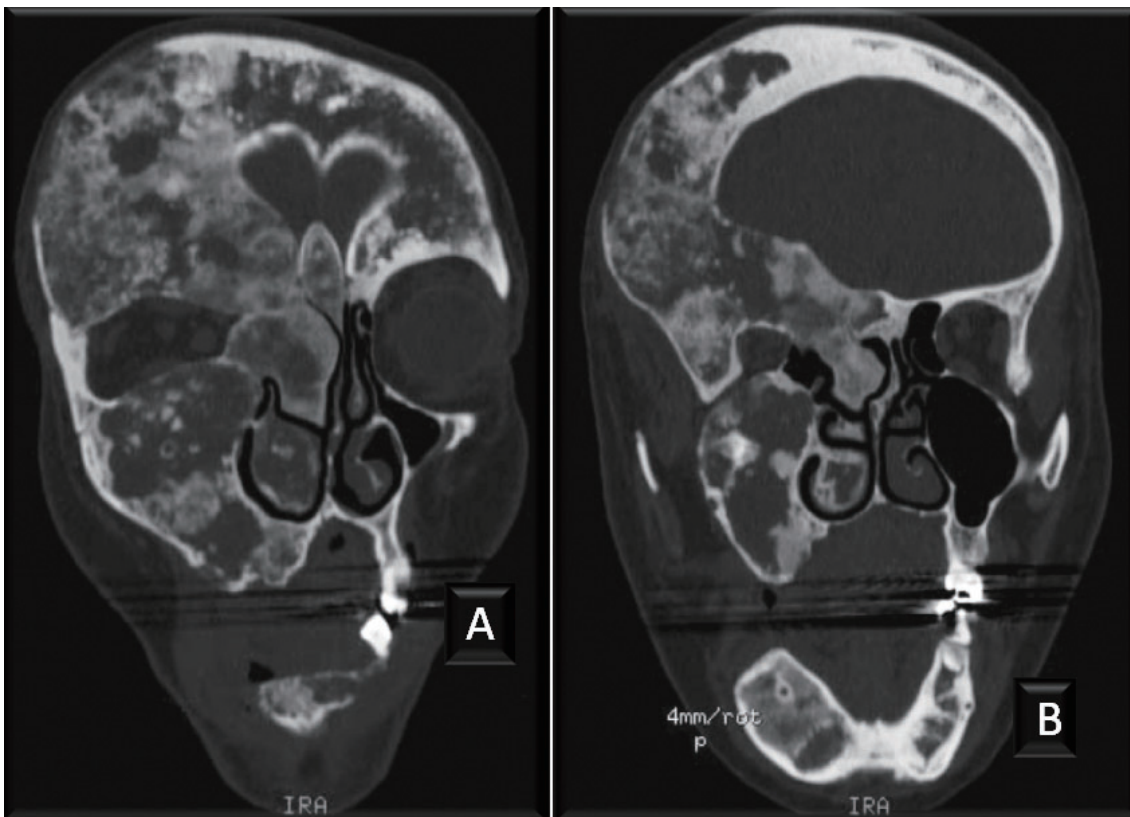


Figure 2. CT scan, bone window and coronal view, showing (A and B) involvement of the Crista Galli, deformity of right orbit, middle and inferior nasal conchae. The right maxillary sinus is totally occupied by bone with a mixed or pagetoid appearance.

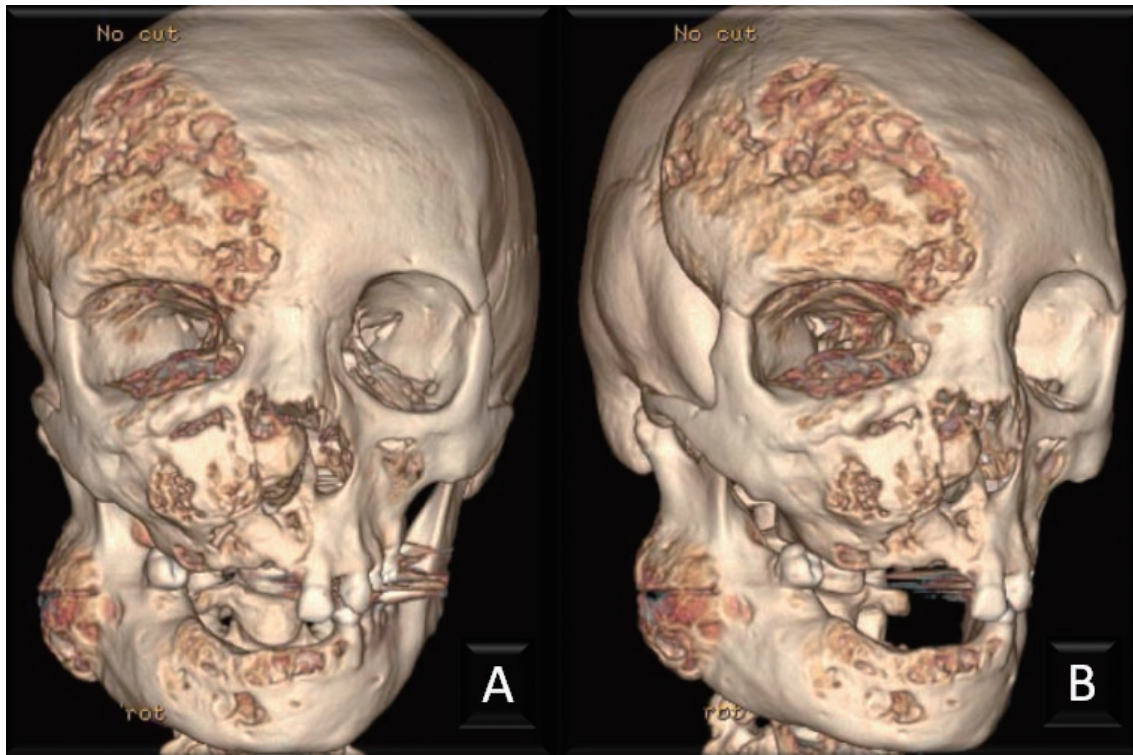


Figure 3. 3D reconstruction shows (A and B) extensive involvement of the right side: frontal and temporal bones, maxilla, and mandible

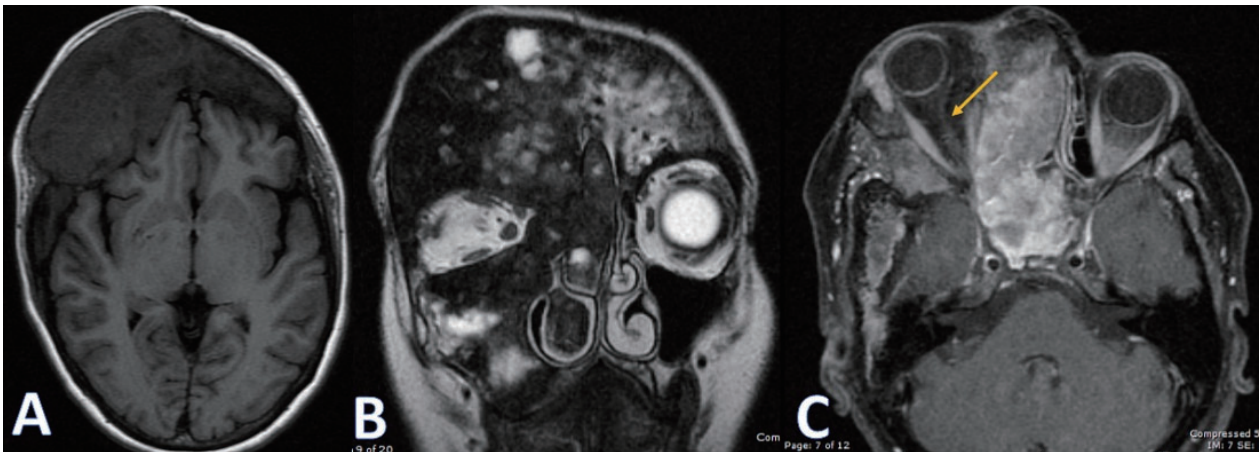


Figure 4. Magnetic resonance imaging (MRI). Large solid expansive craniofacial with proptosis of the right globe and thinness of the optic nerve (yellow arrow). Enhancement post intravenous injection of paramagnetic contrast. A T1 axial view. B T2 coronal view. C T1 fat sat Gadolinium axial view.

Microscopically the lesion revealed of a spread of spindle shaped cells, neoformation of osteoid and bone trabeculae. The trabeculae of woven bone had a variable pattern of distribution, localization and morphology (see Figure 5).

After evaluation, the Head and Neck Cancer Committee recommended periodic control mainly to prevent major vision complications.

3. DISCUSSION

FD is a noninherited rare osseous pathology of unknown origin where the normal bone is superseded by fibro-osseous tissue.^[1,2] FD and McCune-Albright syndrome (MAS) arises from activating missense mutations of the *GNAS* gene, which encodes the α -subunit of the Gs stimulatory protein ($Gs\alpha$). Mutations occur post-zygotically resulting in a mosaic pattern of disease were shown to dramatically upregulate

RANKL expression, consistent with the increased levels of osteoclastogenesis (RANKL promotes osteoclast differentiation and ultimately leads to increased bone resorption).^[3,17] FD represents 3% of all bony tumors and over 7% of all benign osseous tumors.^[4]

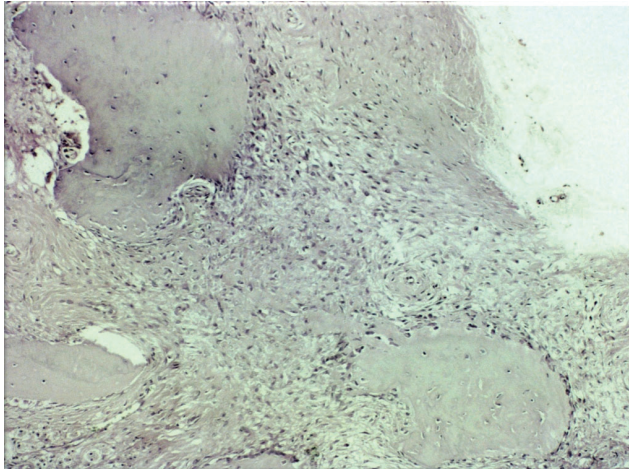


Figure 5. H&E Stain 20×: hypercellular fibrous stroma in which some areas of bone

FD has various clinical presentation groups such as the monostotic, craniofacial and polyostotic forms, and the McCune-Albright syndrome. Monostotic FD involves a single bone and represent 80%-85% of all patients with FD; generally affects the long bones but frequently involves the skull (10%-20% of the patients). The rate of incidence is more highest in the maxilla than in the mandible.^[1,5] Craniofacial FD is restricted to the skull bones and ocular complications, such as visual loss, diplopia, and proptosis, occur in 20%-35% of the cases.^[1] Polyostotic FD affects many bones, is relatively rare, and in some cases, is called the Jaffe type. When it observed, in the polyostotic form, cutaneous pigmentation, no regulated hyperfunction of one or many endocrine glands, we are in presence of the McCune-Albright syndrome.^[1,6,7]

More frequently the FD, was observed in patient between 10 and 30 years old, with equal sex distribution rate; however, the McCune-Albright syndrome is more common in females.^[2]

Microscopically FD shows fibroblast proliferation surrounding islands of woven bone with characteristic look of a “Chinese letters”. The fibroblast show an uniform spindle-shaped nuclei and mitotic figures are not seen.^[5,8]

FD shows variable radiographic features in relation with the level of maturation, which determines the amount of density. Thus, on plain films, FD may display the following

appearances: radiolucent, ground-glass, smoky, cloudy, peau d’orange, finger print, or diffuse sclerosis.^[9,10] A CT bone window displays FD features similar to those exhibited on X ray films, CT attenuation coefficient values, which vary between 34 and 513 Hounsfield units in the different series, in relation with the amount of fibrous and osseous tissue and the rate of bone deposition, leading to three major imaging patterns: lytic or cystic (20%-30%), dense or sclerotic (20%-30%) and mixed or pagetoid (40%-50%).^[11-13]

MRI offers greater specificity in neurovascular and ocular involvement and in detection of other soft tissue lesions. Generally, FD shows an isointense signal on T1 sequence images and a hypointense signal on T2-weighted images. The hypointense signal intensity on T2 sequence images is caused by the osseous deposit. In the first stages of FD, there may be areas of hyperintensity on T2-weighted images.^[10,11,14]

The differential diagnosis of Craniofacial FD must include ossifying fibroma, histiocytosis, Paget’s disease, aneurismal bone cyst, central giant cell granuloma, hemangioma, meningioma, eosinophilic granuloma and brown tumor of hyperparathyroidism.^[6,15] The rate of variation to malignant tumor is more high in monostotic craniofacial disease (0.05%). Transformation to Osteosarcoma, fibrosarcoma and chondrosarcoma are the most prevalent malignant tumors reported in the literature.^[6]

The treatment of craniofacial FD should be focused on the relief of cosmetic and functional problems, which can range from an observational conduct, medical treatment with bisphosphonates, to aggressive surgery. Regular follow-up is required for early detection of disease progression to a malignant transformation.^[16] Studies demonstrated that Denosumab, fully-humanized monoclonal antibody to RANKL recently approved for treatment of osteoporosis and skeletal-related events in adults with bone metastases from solid tumors, was effective for both prevention of lesion expansion and FD-related bone pain.^[17]

4. CONCLUSIONS

Polyostotic craniofacial FD may rarely involve both sides of the cranium, be no symptoms or may present with different neurological signs. The diagnosis depends mainly on the Imaging exams (X ray, CT or CBCT) but clinicians must be aware of the potential diagnosis even if no symptoms are observed. The treatment, in most cases, is conservative, surgery is only indicated in cases with marked facial deformity or neurological complications.

CONFLICTS OF INTEREST DISCLOSURE

Authors declare have no interest conflict.

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