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Craniofacial Impact of Fibrous Bone Dysplasia: Report of a Case and Review of the Literature

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Abstract | **Case Report**

Bone fibrous dysplasia (BFD) is a rare, congenital, sporadic disease of benign bone involvement responsible for bone deformity involving defects in osteoblast dedifferentiation, medullary fiber proliferation and osteoclast hyperactivity. There are two forms of the disease: monostotic, with single lesions, and polysostotic, with multiple lesions. Sarcomatous evolution is exceptional. It may be encountered in syndromic conditions, such as MacCune Albright syndrome, which associates DFO with endocrine disorders and café-au-lait stains; Mazabraud syndrome, corresponding to the association between intramuscular myxoma and DFO. We report a case of DFO collected in the radiology department on August 20, 1953.

Keywords: Fibrous Dysplasia - Bone - CT scan.

INTRODUCION

Fibrous dysplasia is a bone disorder characterized by cortical thinning and replacement of bone marrow by abrasive fibrous tissue containing spicules, resulting in pain, disability and progressive deformity [1].

Bone fibrous dysplasia (BFD) is a benign bone disease, congenital but not transmissible to offspring, in which bone is replaced by fibro-osseous tissue containing immature osteogenesis responsible for fractures, functional disorders, deformity and pain [1]. Sarcomatous evolution is exceptional.

Bone lesions may be single (monostotic form) or multiple (polyostotic form). DFO may be associated with cutaneous macular hyperpigmentation and endocrinopathy as part of McCune-Albright syndrome [2].

The combination of polyostotic DFO and intramuscular myxomas results in Mazabraud syndrome. Bone damage caused by excess FGF-23 leads to urinary phosphate leakage, which may be responsible for hypophosphatemia and osteomalacia.

OBSERVATION

A 35-year-old man with no personal or family history presented with a left frontal swelling of progressive evolution. Questioning revealed notions of headache, chronic left nasal obstruction associated with repeated episodes of epistaxis. The patient had no visual disturbances. The clinical examination was unremarkable.

A CT scan of the brain revealed an expansive bone lesion in the frontal sinus, extending to the left ethmoidal and sphenoidal cells, with a ground-glass appearance and rounded osteocondensation suggestive of fibrous bone dysplasia (Figs 1 and 2).

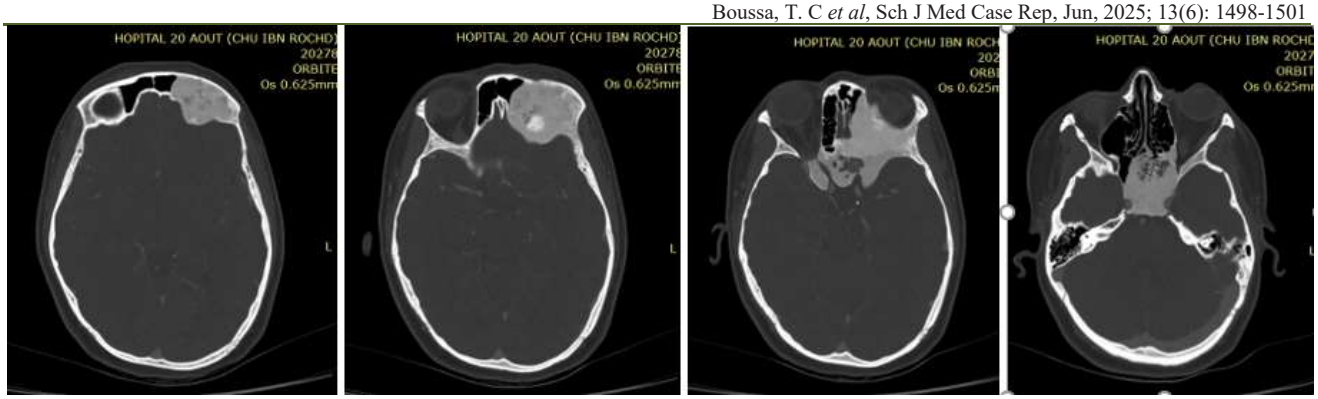

Figure 1: Axial section in bone, expansive bone lesion

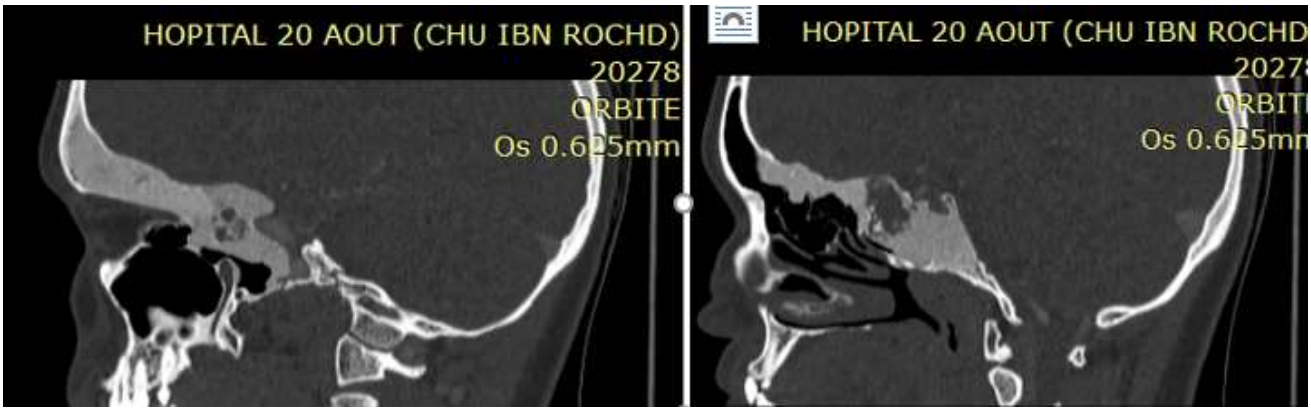

Figure 2: Sagittal bone window cut, expansive bone lesion

DISCUSSION

Fibrous bone dysplasia is a benign skeletal disorder, occurring during infrequent, sporadic, congenital bone growth resulting in deformity.

It results from a mutation in the GNAS gene located on chromosome 20q13 and codes for the subunit of the stimulatory G protein (Gs) occurring at an early stage of embryonic development [3].

Normal bone is replaced by a functionally and structurally abnormal matrix and the marrow spaces show extensive fibrosis with local loss of hematopoiesis.

Its prevalence is less than 1/2000 and underestimated due to asymptomatic forms.

It represents 2.5% of bone lesions and 7% of benign tumor-like bone lesions. It affects both sexes equally and the age at diagnosis is most often between 5 and 30 years old.

The clinical expression of the disease depends on the proliferation, migration and survival of mutated cells during embryonic development.

DFO lesions can be a source of headaches, neuralgia, neurosensory damage (vision, hearing, balance, olfaction), functional disorders (nasal obstruction, dilation of the tear ducts, alignment and dental articulation disorders), infectious complications (sinusitis, otitis, mastoiditis) or being responsible for significant aesthetic damage (occipital, parietal or frontal bumps, leontiasis, prognathism, mandibular damage, exophthalmos, etc.).

They can affect all bones and are either single in the monostotic form (80%), or multiple in the polyostotic form (20%) with a frequently unilateral distribution (metameric or hemimelic) of the affected sites.

Craniofacial damage is present in 50% of polyostotic forms and 27% of monostotic forms [4]. In monostotic forms, maxillary and zygomatic lesions are the most common and those of the temporal bone are rare [5].

Most often, dysplasia lesions have slow, indolent growth. However, in rare cases and before puberty, the lesions may show rapid growth responsible for local complications. In some patients, rapid growth is associated with other pathological lesions such as aneurysmal bone cysts or mucoceles [6-8].

The disease may remain active into adulthood in some patients [9]. Imaging makes the diagnosis when faced with the accidental discovery of a bone lesion.

The clinical examination points towards other bony locations with neurosensory and endocrine damage and the repercussions of this disease on daily life.

The radiological semiology of the lesions is different from one bone to another in the same patient

Standard radiography can highlight three varieties of lesions which can be homogeneous condensing in ground glass; pseudocystic and mixed with a pagetoid appearance [10].

The scanner allows a good semiological characterization of craniofacial fibrous dysplasia lesions with cortico-spongy dedifferentiation, bone thickening, thinned but regular bone tables, possible crossing of sutures and the ground glass appearance with sometimes a cystic or fibrous component.

The particularities of craniofacial lesions are the respect of the cerebral cortex, the narrowing of the foramens, orifices and processions, leading to a neurosensory deficiency.

In the event of endocrine involvement, pituitary MRI can be used to detect a pituitary adenoma or maxillofacial or skull base lesion which may be difficult to distinguish from acromegaly [11, 12].

Bone scintigraphy should be performed in the event of imaging suggestive of DFO without any additional extra-osseous clinical evidence or in the event of an atypical or suspicious lesion of malignancy and allows for mapping of the disease [13].

CT scan coupled with MRI with injection of contrast product are necessary in order to eliminate differential diagnoses (basal plaque meningioma, osteoma, maxillary ossifying fibroma, Paget's disease, etc.), to analyze the extension local and nerve compressions.

In the absence of clinical progression, an annual CT scan for the first two years will allow the assessment of the growth of the lesion (quiescent form without growth, non-aggressive form with low growth and aggressive form with rapid growth). Any change in symptoms requires rapid performance of a CT scan to assess local extension and complications and look for signs of malignant transformation.

CONCLUSION

The craniofacial involvement of craniofacial bone dysplasia is a rare bone pathology which can manifest itself by significant aesthetic and functional disorders. The treatment is interdisciplinary and often poses certain difficulties even after surgery.

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