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Fibrous Dysplasia: Illustrative Case Report

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Abstract **Case Report**

Fibrous dysplasia of bone is a congenital but non-hereditary benign bone disorder in which normal bone is replaced by fibrous tissue containing immature osteogenesis. It is attributed to a mutation of the GNAS 1 gene on chromosome 20q13, an activating mutation of the subunit of the G protein. It is a disease that is most often silent, discovered accidentally on a standard X-ray or revealed by bone pain or a pathological fracture. Imaging and histology, when necessary, help to establish the diagnosis. Although it is not a tumor, it is often classified as a benign bone tumor for reasons of radiographic and anatomopathological differential diagnosis. It may be monostotic or polyostotic or part of McCune-Albright or Mazibraud syndromes. We report here a case of fibrous dysplasia to highlight the contribution of imaging in the positive and also differential diagnosis.

Keywords: Fibrous Dysplasia, imaging, CT.

INTRODUCTION

Fibrous dysplasia is a rare benign congenital bone disease. It causes osteolytic lesions, bone pain, deformities, fractures due to bone fragility and sometimes neurological complications due to nerve compression [1, 2]. This pathology is due to an activating mutation in the of the GNAS 1 gene (guanine nucleotide binding protein, alpha stimulant) affecting somatic cells [3]. It accounts for 2.5% of bone diseases and 7% of bone tumors [4]. Fibrous dysplasia may be monostotic or polyostotic or be part of McCune-Albright or Mazabraud syndromes.

CASE REPORT

Male child, 5 years old, who presented for brain MRI as part of the exploration of his behavioral and social interaction disorders. His MRI revealed a diffuse thickening of the cranial and facial bones, in heterogenous hyposignal T1 et T2, measuring 25mm maximum thickness at the frontal level, with near-total filling of the maxillary, sphenoidal and frontal sinuses, and of the ethmoidal and mastoid cells (Fig 1). There was no cerebral parenchymal abnormality. The patient had an additional CT scan to better analyze the bone, which revealed a ground-glass appearance with diploid enlargement and symmetrical hypertrophy of the facial bones, with near-total filling of all facial sinuses, compatible with craniofacial fibrous dysplasia (fig.2).

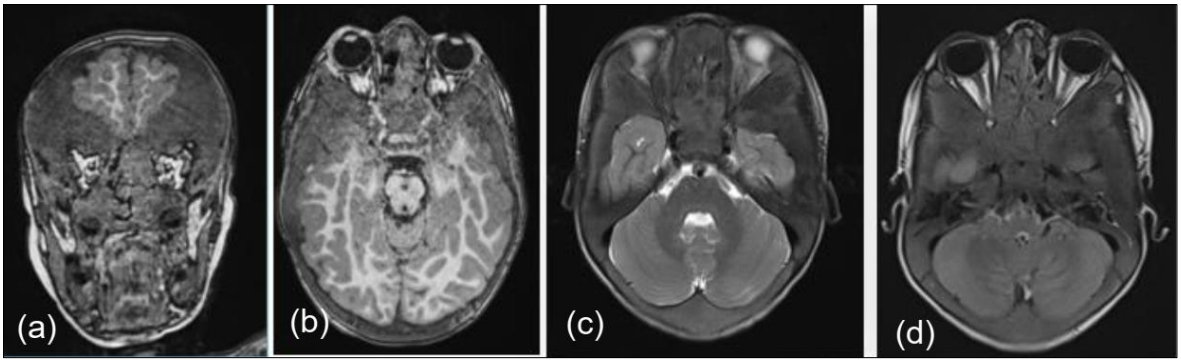

Figure 1: Coronal T1(a), axial T1 (b), axial T2 (c) and axial FLAIR (d) sequences showing heterogeneous thickening of craniofacial bones in the context of fibrous dysplasia

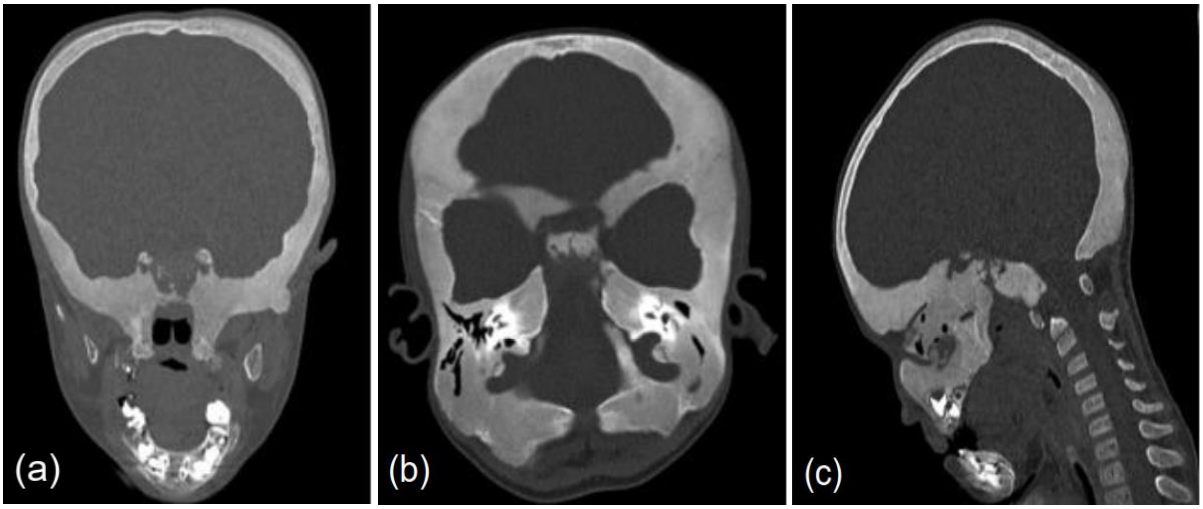

Figure 2: Brain CT-angiography in coronal (a), axial (b) and sagittal (c) sections showing the ground-glass appearance, diploid enlargement and symmetrical hypertrophy of the facial bones, with near-total filling of all facial sinuses, compatible with craniofacial fibrous dysplasia

DISCUSSION

Fibrous dysplasia of bone is a congenital but non-hereditary benign bone disorder in which normal bone is replaced by fibrous tissue containing immature osteogenesis. It is attributed to a mutation of the GNAS 1 gene on chromosome 20q13, an activating mutation of the subunit of the G protein [5]. It is a disease that is most often silent, discovered accidentally on a standard X-ray or revealed by bone pain or a pathological fracture. Imaging and histology, when necessary, help to establish the diagnosis [6-7].

Fibrous dysplasia accounts for 7% of benign tumoral bone lesions, with a prevalence of less than 1/2000, underestimated due to its often-asymptomatic nature [4]. It is not strictly considered as a tumor, even if it is classified as such.

The diagnosis of FD is based on a combination of clinical, biological and, especially, radiological evidence and can be confirmed by anatomopathological study (sometimes).

The average age at diagnosis is between 5 and 30 years [8], with a slight female predominance according to the authors [9]. Lesions most often appear in childhood as in our patient's case and progress very little after puberty. All bones of the body can be affected, and there are monostotic (70%) and polyostotic (30%) [9].

Fibrous bone dysplasia is most often discovered incidentally on imaging, as in our case, or during the work-up of bone pain. Bone lesions can be deforming, hypertrophying or brittle, and can sometimes cause compression of adjacent structures or fractures [9]. Other complications of fibrous dysplasia depend on the localization of the lesion and can vary from simple cosmetic damage to real functional handicap.

The risk of sarcomatous transformation of fibrous dysplasia lesions (osteosarcoma, fibrosarcoma, chondrosarcoma) is relatively low and depends on the shape of the lesion [10].

In typical cases, the diagnosis is easily made because of radiological data alone, although histological confirmation may sometimes be necessary [10].

The radiological aspects of fibrous dysplasia of bone (FD) are varied, reflecting the histological polymorphism of the disease. Thus, the degree of ossification of the tissue will correspond to radiolucent or condensing lesions [11].

Schematically, three types of appearance can be observed: either homogeneous clarity, a smoke-like appearance or a slightly condensed, homogeneous appearance known as ground-glass, as in our case. Homogeneous clarity is the least evocative aspect, while the smoky and ground-glass appearance are suggestive of the diagnosis, since the lesion is too opaque for the size of the lacuna. The frequent presence of lesional calcifications can be found in the periphery or center of lesions. Calcification of cartilaginous islands can simulate a cartilage tumor (enchondroma). The limits of these radiolucent lesions are generally sharp and condensed. The presence of a dense peripheral border is frequent, and immediately orients towards a benign lesion.

Conventional X-rays are usually helpful in making the diagnosis, but CT scans can sometimes be used to aid diagnosis, or to determine the extent of the disease. Specifically, CT can be used to evaluate the disease in different planes, to identify cracks or cortical erosions invisible on plain films [12]. CT can also be useful for evaluating the effect of bisphosphonate treatment on maxillofacial and cranial lesions, with greater ease and accuracy than plain radiography [13].

Overall, the main benefit of CT scans lies in the exploration of cranial and maxillofacial lesions for diagnostic purposes and to search for complications, whereas the for long bones, in which case it will be used to detect erosions or fissures invisible on conventional radiography, in the context of a painful patient with unchanged radiography.

The typical MPO lesion combines some of the following positive signs: lesion of medullary origin, slightly condensed, homogeneous "ground-glass" pattern, highly suggestive; lytic pattern with sharp margins; complete or incomplete osteosclerotic peripheral border; possible presence of intralesional bone trabeculae, creating a compartmentalized appearance; intralesional calcifications, particularly in the proximal femur; thinned and often blown cortex; deformity, bone hypertrophy. Negative signs of malignancy, such as cortical lysis, periosteal reaction or extension to the soft tissues, must be ruled out [10].

DFO can take on different CT appearances, which may be associated within the same lesion or on different lesions in the same patient [14].

The MRI appearance of the DF varies according to the degree of mineralization and the histological nature. In spin-echo T1, DF presents as a homogeneous moderate hyposignal, whereas the signal varies considerably on T2. In two-thirds of cases, the lesion will be hypersignal [14]. The intensity of the T2 signal actually depends on the degree of intralesional mineralization. The highly mineralized foci are marked by an intense hyposignal on both sequences. The diagnosis is facilitated by the presence of a border in frank hyposignal, which separates the fibrous tissue from the adjacent normal bone.

Fibrous dyplasia lesions should not change significantly in radiological appearance after puberty; if they do, a differential diagnosis or radiological appearance after puberty; if they do, a differential diagnosis or malignant transformation should be considered [15].

Particularities

If a fibrous dysplasia lesion is found, it may be interesting to analyze the soft tissues in the proximity of the lesion in search of a soft tissue myxoma as part of a possible Mazabraud syndrome [16]. The Mazabraud syndrome is associated with DF and intramuscular myxomas, generally located in the proximity of bone.

McCune-Albright syndrome combines precocious puberty, polyostotic fibrous dysplasia, skin patches and endocrine disorders such as thyroid nodules with hyperthyroidism, adrenal hyperplasia with hypercorticism, pituitary tumors with acromegaly or hyperprolactinemia [16].

CONCLUSION

Fibrous dysplasia is a benign pathology that's easy to detect if we're familiar with its radiological features: the lesion is intramedullary in location, the "ground-glass" appearance is highly specific, and is frequently associated with a lytic component and peripheral osteosclerotic edging.

The topography of the lesions is also a strong argument: the costal grill, skull or femur are classic for the monostotic form, and the unilateral hemimelic distribution is highly suggestive of the polyostotic form. Cases of doubtful appearance or location require biopsy.

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