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CASE REPORT

TRIPLE LOCALIZED CRANIO-FACIAL FIBROUS DYSPLASIA: A CASE REPORT

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ABSTRACT:
Introduction: fibrous dysplasia of bones is a non-hereditary congenital benign bone disorder, where normal bone is replaced by pseudofibrous tissue containing immature osteogenesis.
Case report: a 29-year-old patient with chronic hemodialysis who had a swollen mouth and hard palate that had been evolving for a year, impeding chewing and swallowing and causing facial asymmetry. Cranio-facial CT revealed multiple osteolytic bone-blast lesions, the histopathological study of which favored polyostotic fibrous dysplasia.
Discussion: fibrous dysplasia lesions may be single or multiple and may be responsible for pain and fragility, causing neurological complications in craniofacial localization. Imaging and, when a biopsy is needed, histology can establish the diagnosis. The treatment is based on bisphosphonates or, in special cases, surgical excision.
KEY WORDS: fibrous dysplasia, polyostotic, craniofacial.

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INTRODUCTION

Fibrous dysplasia (FD) is a rare congenital disorder; it may be single in the monostotic form or multiple in the polyostotic form [1]. It is a disabling disease that causes fractures, pain, deformities and functional impotence [2]. FD is due to a postzygotic mutation of the Gnas1 gene responsible for immature osteogenesis in a bone weakened by fibrosis [3]. This mutation also affects other cell types; it can be associated with cutaneous manifestations ("café au lait" stains) and endocrine (early puberty, hyperthyroidism, acromegaly, Cushing's syndrome) in the context of McCune-Albright syndrome, or with myxomas in Mazabraud syndrome [3]. Cranio-facial lesions are present in 50% of polyostotic forms and 27% of monostotic forms [2]. It is a pathology that is most often silent, discovered accidentaly on a standard radiography or revealed by a bone pain or a pathological fracture. Imaging and histology, when necessary, provide the diagnosis. There are many reports of medical treatment with bisphosphonates showing a decrease in lesion pain but no resolution of the growth itself. Surgical treatment is variable depending on the location of the lesion and the symptoms. We report the case of a polyostotic FD in chronic hemodialysis with craniofacial involvement.

CASE REPORT

Mrs. B.L, aged 29, followed for 8 years for chronic renal failure at the hemodialysis stage on indeterminate nephropathy, who consulted for a tumefaction of the buccal floor and the hard palate evolving since a year, hindering the mastication and the swallowing and causing right eye pain. The clinical examination objectified three masses. The first sitting on the anterolateral part of the buccal floor, at the expense of the symphyseal internal cortical, well circumscribed, between the 32 and the 46th teeth (fig1 A). The second was a localized mass at the level of the hard palate, bilobed, well-defined extending posteriorly to the osteo-membranous junction of the palate, leading to a decrease in nasal flow on the right side (fig1 B) and the third in front of the rising branch of the right mandibular bone, which is responsible for obvious facial asymmetry.

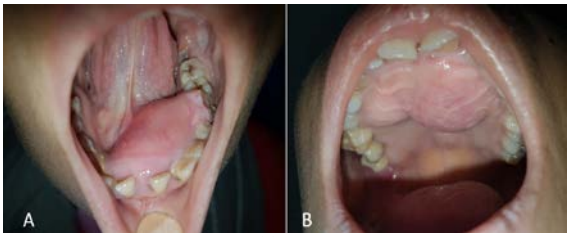

Fig 1: Preoperative photograph of fibrous dysplasia lesions located in the buccal floor (A) and hard palate (B)

The three masses were hard, painless, adherent to the deep plane, covered with a healthy mucous membrane. The neurological examination did not reveal any deficit signs. The biological test revealed a hyperparathyroidism at 2416 pg / ml (N: 15-65 pg / ml) with a high level of alkaline phosphatase at 508 IU / L (N: 40-150 IU / L), without other disturbances in the phosphocalcic balance. A cranio-facial CT revealed multiple osteolytic lesions of the right maxillary bone, the buccal floor and the right mandibular angle, blowing the bone especially around the maxillary sinus, with an overall osteo-condensed appearance of the cranial vault (fig2).

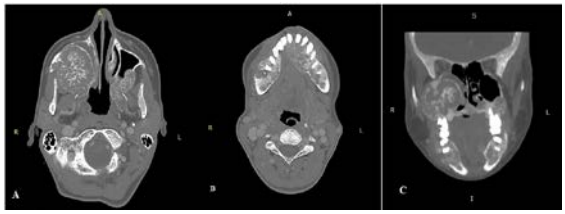

Fig 2: Pre-operative CT images shows multiple osteolytic lesions of the right maxillary bone, the buccal floor and the right mandibular angle, blowing the bone especially around the maxillary sinus (A and B: axial section, C: coronal section)

A block excision of the right maxillary lesion by transpalatine and para-latero-nasal approches was conceived, as for the lesion of the oral floor, we proceed by endobuccal way preserving the excretory channel of Wharton and the lingual pedicle (fig3).

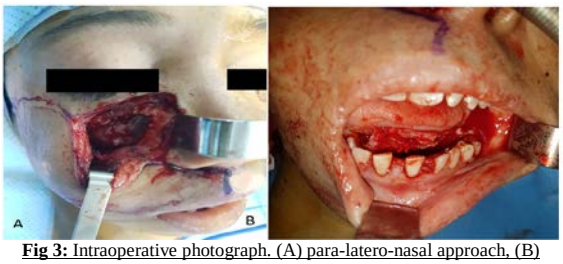

Fig 3: Intraoperative photograph. (A) para-latero-nasal approach, (B) Endocuccal surgical approach

The anatomopathological study revealed a tissue of tumor shape, with a benign conjunctival proliferation, made of fibroblastic cells arranged in diffuse layers with sometimes rounded bone spans, sometimes elongated in "Chinese letter" and some giant osteoclastic cells, corresponding to fibrous dysplasia. Postoperative follow-up was simple with disappearance of respiratory signs and masticatory problems and improvement of swallowing (fig4).

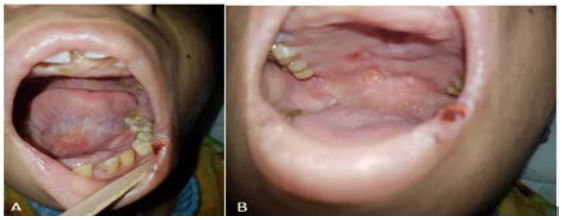

Fig4 (A) and (B): Post-operative aspect of the patient after excision of the lesions

In a second step, the patient benefited from a subtotal parathyroidectomy. The evolution was marked by the regression of hyperparathyroidism.

DISCUSSION

Fibrous dysplasia is a rare developmental disorder in which normal bone is replaced by fibrous tissue [4]. It was originally described in 1891 by Von Recklinghausen. The literature was enriched later by several case reports [5]. It is due to a mutation of the GNAS 1 gene, this mutation activates the α subunit of the G protein, which occurs mainly in the R201 position, which includes the intrinsic GTPase domain of the molecule. This results in stimulation of the adenylate cyclase, and overproduction of cAMP and then overexpression of the c-fos protein which is at the origin of a lack of differentiation of the osteoblasts [6]. It is indeed a rare non-hereditary congenital disorder of chance discovery or as a result of complications. The estimate of its prevalence is less than 1 in 2000 but remains difficult to determine given the frequency of asymptomatic forms [1]. It is divided into two clinical types; we distinguish the monostotic form and the polystolic form. It represents 7% of all benign bone tumors. Men and women are equally affected. Most cases are diagnosed before the age of 30 [3]. The craniofacial region is affected in 25% of cases; the most affected areas are the maxillary bone, the mandible, the ethmoid bone and the frontal bone [2].

Cranio-facial FD lesions may be asymptomatic or may be revealed by headache, neuralgia, neurosensory or functional disorders, infectious complications or significant aesthetic damage [7], as was the case of our patient who presented facial asymmetry induced by the large mass that developed at the level of the two bones: mandibular and maxillary right. The diagnosis is confirmed most often by the radiological findings which will highlight an osteolytic lesion, often heterogeneous, mainly radio-transparent, with a thin cortex, sometimes a bone hypertrophy, with often a border of osteocondensation peripheral to the lesion, but with, in some places, bone condensation called "frosted glass" very suggestive of the diagnosis [8]. CT shows bone thickening, cortico-cancellous dedifferentiation, thinned but regular bone tables, the possible crossing of sutures and the frosted glass appearance, sometimes with a cystic or fibrous component. The particularities of the craniofacial involvement are the respect of the cerebral cortex, which can be repressed, and the narrowing of the foramina, orifices and parades, responsible for neurosensory disorders [1]. Bone scintigraphy is performed at the beginning of the treatment to detect all the bone lesions of the FD, the dysplastic lesions are hyperfixing [1]. MRI has no value in the diagnosis of FD, but only if there is suspicion of malignant transformation that is rare and seen only in 0.4 to 4% of cases. Osteosarcoma is the most common but fibrosarcoma or chondrosarcoma can also be seen [4]. Bone remodeling markers are used to evaluate disease activity and response to treatment. In practice, a minimum of tests must be carried out in front of a FD to evaluate its activity and to look for phosphorous diabetes. PAL (total / bone) assay is a good way to monitor disease activity as it is increased in almost 75% of patients [7]. The histological study, when it is done, makes it possible to demonstrate fibrous focal proliferation within the bone tissue, made of preosteoblastic cells anarchically producing an immature bone matrix. It was shown by histomorphometric analysis performed on bone biopsies in lesional tissue that there was a clear lesion of secondary hyperparathyroidism in dysplastic tissue [3]. As a result,

several reported cases of FD in the literature were associated with hyperparathyroidism, most often secondary, including my own.

There are two clinical forms in FD: McCune-Albright syndrome, which combines precocious puberty, polyostotic FD, skin-coffee stains and endocrine abnormalities such as thyroid nodules with hyperthyroidism, adrenal hyperplasia with hypercorticism, pituitary tumors with acromegaly or hyperprolactinemia. And Mazabraud syndrome, which associates FD and intramuscular myxomas, which are generally close to bone lesions, and tend to recur after surgical excision [7].

The therapeutic management of FD consists of two components: medical treatment and surgical treatment. The goal of treatment is to improve the quality of life of patients by reducing pain, controlling the activity of the disease, and preventing and managing its complications [9]. The medical treatment is based on the use of anti-resorptive drugs such as pamidronate which is the cornerstone of the available treatments, used at the dose of 60 mg / day for 3 days with a cure every 6 months. This treatment reduces pain, improves radiological appearance and bone mineral density, but has only been evaluated in open studies. Supplementation with calcium (1 g / d) and vitamin D3 (800 IU / d) is strongly recommended, given the frequency of vitamin D deficiency seen in patients with FD. In addition, poorly differentiated osteoblastic cells have been shown to produce an excess of interleukin-6 (IL-6), which inhibits bone formation and stimulates osteoclast formation, and RANKL, receptor activator of NF membrane receptor ligand, -kB, which stimulates osteoclastogenesis and bone resorption. Several clinical cases have shown a beneficial effect of tocilizumab, a monoclonal antibody directed against the IL-6 receptor, and denosumab, a monoclonal antibody directed against RANKL, with patients who do not respond to bisphosphonates [10].

Surgical treatment is essential in case of pain that is resistant to medical treatment, functional damage (the case of our patient) and esthetics with a demand for correction, or progression of the lesions, especially when they may reach the base of the patient skull or the eyeball [10]. with our patient, we performed a resection surgery, consisting of a total excision of the masses of the maxillary bone and the floor of the mouth with preservation of adjacent anatomical structures and aesthetic damage.

CONCLUSION

FD is a benign bone disease but can be serious and debilitating that. We must think of in case of headaches, neuralgia, sensory impairment, functional disorders, infectious complications (sinusitis, otitis, mastoiditis). The diagnosis is based on imaging data, in particular CT. In case of pain, medical treatment relies on bisphosphonates. Surgical resection is indicated only if the disease is symptomatic, especially in case of neurosensory disorders, or for aesthetic purposes. Patient monitoring should be extended, the prognosis remaining related to the risk of sarcomatous degeneration of the lesions.

PATIENT CONSENT

Written informed consent was obtained from the patient for publication of this case report.

COMPETING INTERESTS

The authors declare no competing interests.

AUTHORS' CONTRIBUTIONS

The participation of each author corresponds to the criteria of authorship and contributorship emphasized in the Recommendations for the Conduct, Reporting, Editing, and Publication of Scholarly work in Medical Journals of the International Committee of Medical Journal Editors. Indeed, all the authors have actively participated in the redaction, the revision of the manuscript and provided approval for this final revised version.

REFERENCES

[1] [1] Couturier A, et al. La dysplasie fibreuse osseuse cranio-faciale. Rev Med Interne (2016), http://dx.doi.org/10.1016/j.revmed.2016.02.008

[2] [2] Couturier A, et al. Craniofacial fibrous dysplasia: A 10-case series. European Annals of Otorhinolaryngology, Head and Neck diseases (2017), http://dx.doi.org/10.1016/j.anorl.2017.02.004

[3] [3] Hanifi B, et al, Craniofacial fibrous dysplasia, Clin Imaging (2013), http://dx.doi.org/10.1016/j.clinimag.2013.01.010

[4] [4] A.S. Godse et al. Fibrous dysplasia of the maxilla. Journal Of Pediatric Surgery. (2009) 44, 849–851.

[5] [5] Chapurlat RD, Orcel P. Fibrous dysplasia of bone and McCune- Albright syndrome. Best Pract Res ClinRheumatol. 2008; 22(1):55- 69.

[6] [6] Turan S, Bastepe M. GNAS Spectrum of Disorders. Curr Osteoporos Rep. 2015 Apr 8.

[7] [7] N. Akasbi et al. La dysplasie fibreuse : état des lieux. Pan African Medical Journal. 2015; 21:21 doi:10.11604/pamj.2015.21.21.3487

[8] [8] Gensburger .D, Chapurlat. R.D, Rey-Jouvin .C, Orcel. P. Prise en charge de la dysplasie fibreuse : recommandations actuelles et perspectives thérapeutiques. La Lettre du Rhumatologue. No 409-février 2015.

[9] [9] J. Numminen et al. Polyostotic fibrous dysplasia of the mandible and bony nasal septum with intracranial expansion. Egyptian Journal of Ear, Nose, Throat and Allied Sciences (2016) 17, 49–51

[10] [10] E. Béquignon et al. Craniofacial fibrous dysplasia surgery: A functional approach. European Annals of Otorhinolaryngology, Head and Neck Diseases. Volume 130, Issue 4, September 2013, Pages 215-220