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Case report / Caso clínico

Fibrous bone dysplasia: A case report

Displasia fibrosa ósea: reporte de caso

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ABSTRACT

Introduction: Fibrous bone dysplasia is a bone disease that affects the normal composition of the bone, in any part of the skeletal system, whether it is monostotic or polyostic, causing tumor masses of fibrous connective tissue. At the same time, it encompasses a wide phenotypic spectrum that can vary depending on the age of onset and the affected apparatus, such as endocrinopathies. Regarding its diagnosis, if it only affects a single bone without any other finding, it would only need histopathological confirmation. For its treatment could be surgery, antiresorptive drugs such as zolendronic acid, immunological medications and pain management if required. **Clinical Case:** A 14-year-old boy presents with facial asymmetry in the Internal Medicine department. Diagnostic workshop: In computed axial tomography of the skull, an expansive sclerotic lesion with ground glass density is evident at the level of the upper jaw and right zygomatic region. The pathology report of a biopsy taken confirmed the diagnosis of fibrous bone dysplasia. **Conclusion:** This type of dysplasia is rare, and may have a non-progressive asymptomatic course, or cosmetic changes and exacerbating pain in the progressive stage, which poses a challenge for the patient's diagnosis. Several factors must be taken into consideration to choose the best treatment for the patient. For this reason, it should be studied early for an early therapeutic decision.

Keywords: Fibrous dysplasia of bone, monostotic, maxillectomy, zoledronic acid.

RESUMEN

Introducción: La displasia fibrosa ósea es una enfermedad que afecta la composición normal del hueso, en cualquier parte del sistema esquelético, ya sea de forma monostótica o polióstica, y provoca masas tumorales de tejido fibroso conectivo. A su vez, engloba un amplio espectro fenotípico que puede variar según la edad de aparición y aparatos afectos como las endocrinopatías. En cuanto a su diagnóstico, en caso de que solo afecte a un solo hueso sin ningún otro hallazgo, se necesitaría únicamente confirmación histopatológica. Su tratamiento se basa en la cirugía, fármacos antirresortivos, como el ácido zolendrónico, medicamentos inmunológicos y manejo del dolor en caso de que se requiera. **Caso clínico:** Un adolescente de 14 años acude al servicio de Medicina Interna con asimetría facial. En la tomografía axial computarizada de cráneo, se evidencia una lesión expansiva esclerótica con densidad de vidrio esmerilado en el maxilar superior y la región cigomática derecha. El reporte de patología de una biopsia confirmó el diagnóstico de displasia fibrosa ósea. **Conclusión:** Este tipo de displasia es infrecuente y puede tener un curso asintomático no progresivo, o también presentar cambios cosméticos y dolor exacerbante en la etapa progresiva, lo que representa un desafío en el diagnóstico del paciente. Varios factores deben tomarse en consideración para elegir el mejor tratamiento para el paciente. Por tal motivo, debe estudiarse de manera temprana para su decisión terapéutica oportuna.

Palabras Clave: Displasia fibrosa ósea, displasia fibrosa monostótica, maxilectomía, ácido zolendrónico.

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1. Introduction

Fibrous bone dysplasia is a rare, benign, and congenital pathology that affects the skeletal development of the human body by altering the normal composition of the bone [1]. It has a global prevalence of 1/100 000 inhabitants and comprises 5% of all primary bone tumors [2]. Although its etiology is not clear, post-zygomatic mutations of the GNAS gene, located on chromosome 20, have been identified. They lead to the stimulation of adenylyl cyclase and subsequent overproduction of cyclic adenosine monophosphate with uncontrolled cell proliferation and inadequate differentiation [1,3]. Its clinical spectrum is broad, it could be accompanied by dermatological or endocrinological disorders. The diagnosis is made through clinical history, radiological studies depending on the affected bone, and histopathological investigation. Management focuses on observation, pharmacological use with bisphosphonates and surgical treatment [4,5]. There are very few reports in national medical literature on this benign pathology, which is why its scientific dissemination is considered essential.

2. Clinical case

A 14-year-old male patient arrived at the internal medicine department due to facial asymmetry with size increase of the right side of the face for approximately 5 years, without any other symptoms. He denies any personal pathological or surgical history, with a complete vaccination schedule. Physical examination revealed the previously described facial asymmetry, harmony of the upper and lower extremities, and Tanner Score according to age. The rest of the examination shows no significant findings.

In a tomography of the skull and paranasal sinuses (Figure 1), an expansive sclerotic lesion with ground glass density is evident at the level of the upper jaw and right zygomatic region, which remodels and reduces the size of the respective paranasal sinus.

A radiological series is performed on both hands, backbone, lumbosacral, pelvis and femur with a bone age according to that of the patient, without pathological findings.

In laboratory studies, parathyroid hormone 59.3, LH 4.12, FT4 1.32, TSH 2.93, testosterone 6.12 and vitamin D 28.38 were reported; thus, he received vitamin D3 40 000 IU for 2 weeks and zolendronic acid 4 mg intravenously in a single dose.

An excision of the benign tumor and a Cadwell biopsy of the right jaw was performed. Its microscopic study confirmed the diagnosis of fibrous bone dysplasia.

He was intervened for a right maxillectomy, mesh placement under 3D planning and reconstruction with a temporal flap, without complications. The pathology report revealed a homogeneous whitish-brown mass measuring 7.2 x 5.4 x 4.4 cm and confirmed the previous diagnosis.

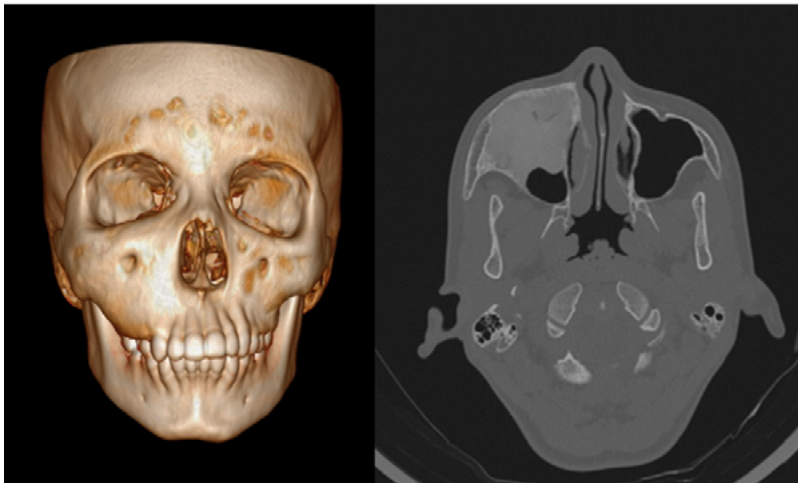

Figure 1. 3D skull tomography and axial tomography of the paranasal sinuses with bone window. Source: SOLCA Hospital – Guayaquil.

Twelve days after the surgical procedure, a new tomographic study of the face (Figure 2) was performed, in which post-surgical inflammatory changes were identified at the level of the surgical bed that extended to the lateral edge and floor of the orbit, as well as the right lateral wall of the orbit, nasal cavity and ipsilateral temporal-parietal region, without evidence of residual lesion and/or recurrence of the tumor.

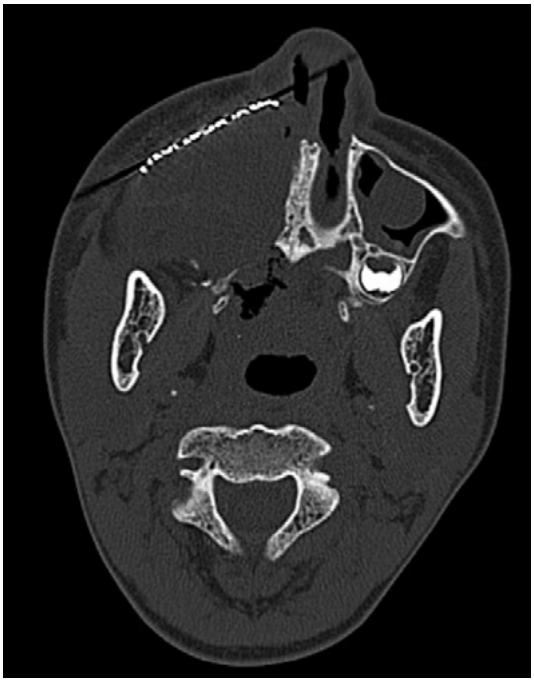

Figure 2. Axial tomography with contrast 12 days after surgery.
Source: SOLCA Hospital – Guayaquil.

Sixteen days after surgery, the patient was evaluated in the outpatient clinic, showing improvement of the facial asymmetry, with a favorable condition.

3. Discussion

Fibrous bone dysplasia is a rare pathology with a wide phenotypic and multi-organ spectrum, whose diagnosis is often challenging in the Internal Medicine field. Also, this condition can affect a single bone (monostotic, as in this case), or several (polyostotic). The former is more prevalent in 70-80% of cases [6,7]. There is no data that determines the prevalence of this disease in Ecuador [8].

Extraosseous manifestations might be associated with café-au-lait macules, hyperthyroidism, acromegaly/gigantism, and abnormal production of testosterone or estrogen, as defined in McCune-Albright syndrome [9,10]; or coexist with intramuscular myxomas in Mazabraud syndrome [1]. During the physical and complementary examinations of this case, none of these alterations were evident.

The diagnostic confirmation of the disease is anatomical-pathological, more reliable for monostotic conditions, since there are several differential diagnoses such as cancer, simple bone cyst, Paget's disease, ossifying fibroma, and giant cell granuloma [1,5,11]. Its malignant transformation is very rare in this type of dysplasia (between 0.4 to 4%) and could help determine polyostotic conditions [12].

Surgery is one of the main treatment options considering the site of the condition and the characteristics of the patient [9]. Various indications for this procedure in craniofacial conditions include compressive neuropathies (such as the optic nerve), severe malocclusion, bone pain resistant to analgesic treatment, high risk of recurrence of the deformity and cosmetic purposes [4,8,13].

Other therapeutic options are bisphosphonates due to their inhibition of bone resorption and decrease of osteoclast production; however, they did not show improvement in radiological findings or in the prevention of expansion of bone lesions [14,15]. Despite this, some intravenous formulations, such as zolendronic acid or pamidronate, have a better therapeutic role compared to the oral route in the treatment of pain related to bone fibrosis [14,16]. Prior to using antiresorptive drugs, it is important to determine the levels of kidney function and calcium phosphorus metabolism (including vitamin D and parathyroid hormone), in case it is necessary to supplement due to deficiencies [3].

Apart from the use of bisphosphonates, another choice includes anti-RANKL antibodies (denosumab) and anti-IL6, which inhibit bone proliferation and reduce pain symptoms, respectively, in case bisphosphonates do not have good efficacy or are contraindicated [15,17].

4. Conclusions

Fibrous bone dysplasia is a benign and rare disease characterized by abnormal growth of bone tissue, which may be accompanied by dermatological and/or endocrinological alterations. Early diagnosis allows for more exhaustive studies and more effective therapy and relief for the patient, highlighting surgical resolution as the first step.

5. Administrative information

5.1 Additional files

None declared by the authors

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5.3. Author contributions

Noemi Bautista Litardo: Conceptualization, methodology, formal analysis, research, project administration, writing of the original draft. **Raúl Peralta Rodríguez:** Conceptualization, Methodology, Research, visualization, writing – review and editing. Both authors read and approved the final version of the manuscript.

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None

5.5. Availability of data and materials

Data are available upon request to the corresponding author. No other materials were reported

5.6. Statements

5.6.1. Ethics committee approval

Clinical cases were not needed

5.6.2. Declaration

The manuscript has not been previously published, nor is it currently under editorial review for publication in another journal.

5.6.3. Consent for publication

The patient's legal guardian provided written consent for the publication of this clinical case.

5.6.4. Conflicts of interest

The authors declare no conflict of competence or interest.

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