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Clinical and imaginological manifestations of cherubism: A case report

Manifestações clínicas e imaginológicas do querubismo: Um relato de caso

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ABSTRACT

Introduction: Cherubism is a rare hereditary disorder characterized by bilateral enlargement of the gnathic bones in children, with a tendency to spontaneous remission at puberty. In some cases, it can develop without a family history. Objective: This report presents a case of non-hereditary cherubism affecting dentition in a 7-year-old patient. Case report: A 7-year-old male patient complained of bilateral swelling of the face in the region of the body and mandibular ramus, with a 3-year evolution, asymptomatic and slow growth, with no history of similar cases in the family. Intraorally, the increase in volume extended from canine to molar, bilaterally, with anomalies in the teeth involved. Hematological tests were within limits. Panoramic radiography and CT scans of the face revealed multiple multiloculated lesions in the body and mandibular branch bilaterally. Incisional biopsy was performed under local anesthesia, which revealed the presence of multinucleated giant cells. Based on the clinical, imaging and histopathological findings, the final diagnosis was cherubism. Due to the self-limiting behavior of the pathology, surgical interventions were not necessary and the case will be monitored. Discussion: Although the lesions tend to be symmetrical, here, they were more prominent on the right side. Radiographic features were typical of the disease. The lesions can cause malocclusions due to ectopic eruptions, as seen in this case. Despite the hereditary characteristic, there was no family history of the disease. Histological and laboratory analyses are important to rule out other fibroskeletal lesions, but clinical and radiographic aspects are paramount in the diagnosis. Upon reaching bone maturation, the lesion tends to regress. However, cosmetic surgery may still be needed later. Conclusion: The diagnosis of the disease should be based mainly on clinical and imaging findings, since histological analysis has limited value. In view of the confirmation, the case should be followed up and surgical interventions postponed if they are necessary later.

Keywords: Cherubism, Mandible, Oral pathology, Clinical diagnosis, Diagnostic imaging.

INTRODUCTION

The face, perhaps more than any other part of the body, has a unique role in our social life [1]. It acts as our "business card", our main reference in our social relationships. Although this

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social function is often overlooked, when something affects facial aesthetics, the diagnostic and therapeutic role tends to be the function of the dental community. First described in 1993 as a "multilocular genetic disease of the gnathic bones," cherubism got its nickname because of the characteristic rounding of the face caused by hypertrophy of the bones, reminiscent of the cherubic angels of Renaissance paintings [1, 3, 4, 5].

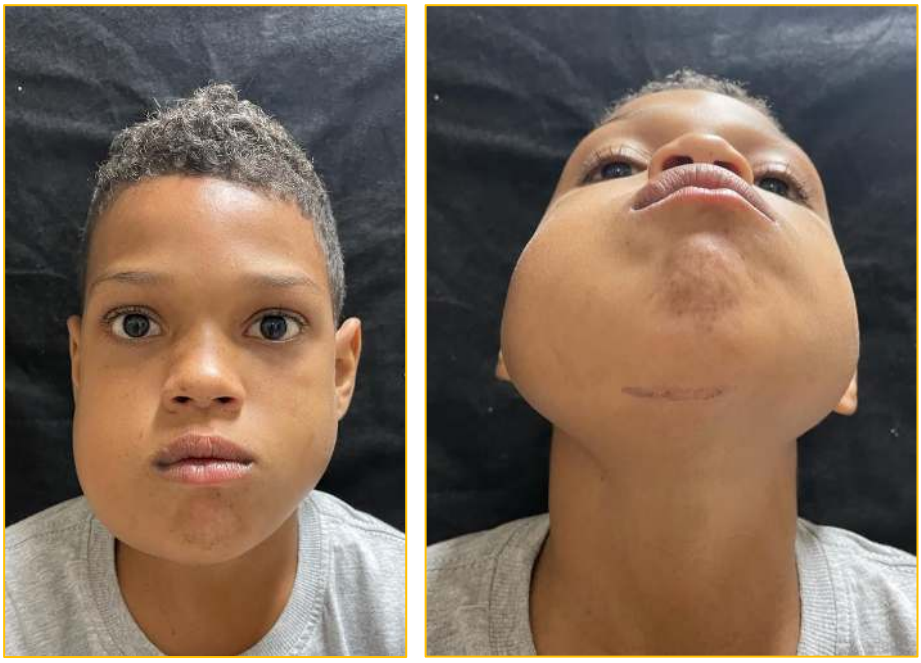
Although the condition was initially characterized as genetic, non-hereditary and sporadic cases have been described in the literature 1,7. Due to the rarity of this entity, it is difficult to determine its prevalence in the population, due to the limited number of cases in the literature 2,4. The sample of non-hereditary cases is even smaller. While its familial form is inherited by an autosomal dominant mutation, its non-hereditary origin is assumed to be caused by new genetic mutations or genetic heterogenicity (mutations in more than one gene) [2].

Cherubism is a type of benign bone dysplasia that affects children and results in bilateral progressive volume enlargement in one or both gnathic bones, with a typically multilocular radiographic presentation [2,3,4]. Although the condition tends to regress spontaneously at puberty, surgical interventions may be necessary for aesthetic or functional reasons [3, 4]. This case report presents a non-hereditary manifestation of cherubism in a 7-year-old patient with bilateral mandibular enlargement affecting his dentition.

CASE REPORT

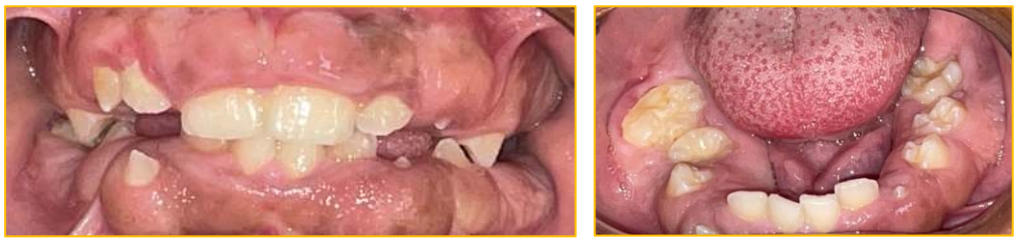
A 7-year-old male patient presented to the Oral and Maxillofacial Surgery and Traumatology Outpatient Clinic (CTBMF) of the Roberto Santos General Hospital with the main complaint of bilateral swelling of the face with a 3-year evolution. The increase in volume was asymptomatic, slow-growing, and there was no history of similar cases in the family. The patient was continuously using anticonvulsant medication (carbamazepine 20mg/ml) to control seizures. Extraoral physical examination revealed a bilateral increase in volume in the region of the ramus and mandibular body, with greater prominence on the right side. The overlying skin was normostained, with no floating point.

Figures 1 and 2: Patient presenting with volume increase in the lower third of the face bilaterally with prominence on the right side



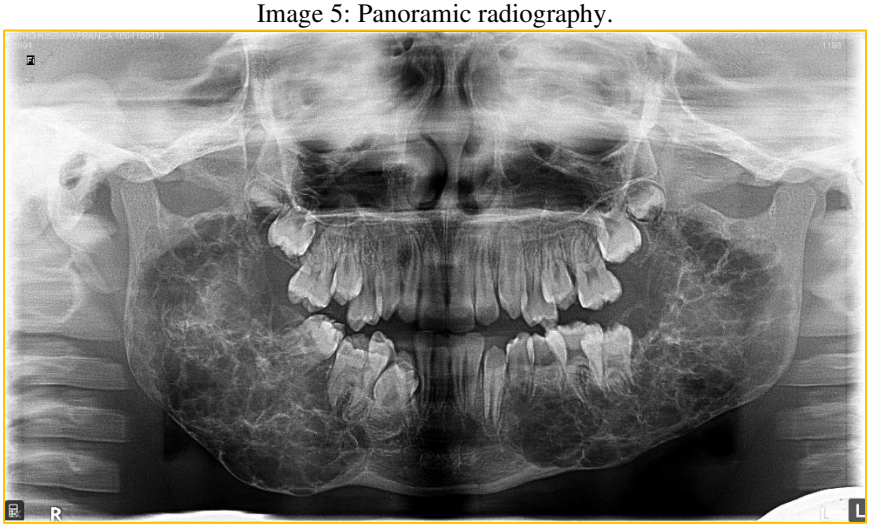
Intraoral examination revealed an expansion of the vestibular and lingual corticals, which extended from canine to molar, bilaterally, with anomalies in the teeth involved. Despite the anomalies, there were no complaints of difficulty chewing. The teeth were normal in size, but had disorders of eruption, shape and positioning in the arch. The patient has mixed dentition, other mucous membranes and satisfactory oral hygiene

Figures 3 and 4: Lingual and vestibular cortical expansion of the mandible extending from the canine region to the molars

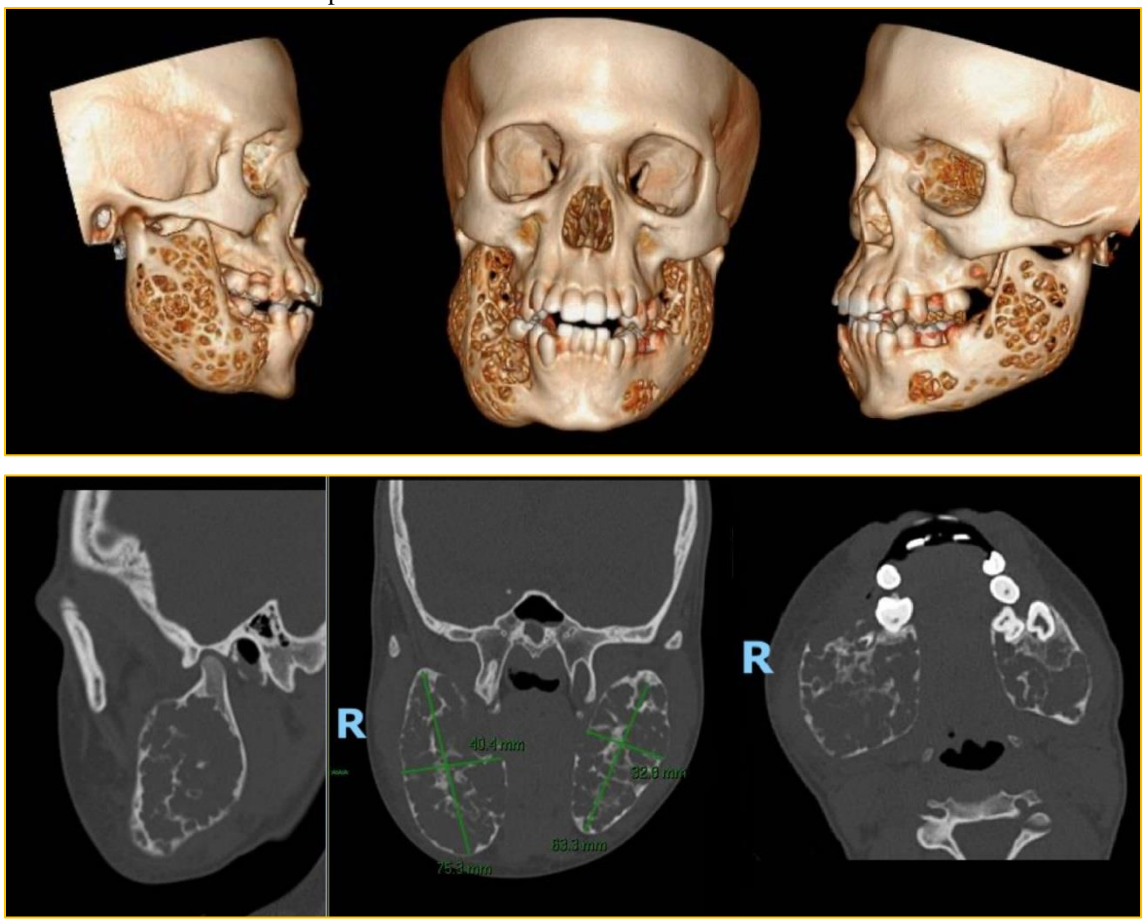


Panoramic X-ray revealed multiple multiloculated lesions in the mandibular body and ramus bilaterally up to its basal base, but not involving the mandibular condyles, tooth displacements with aspects of floating teeth, but without resorption of their roots (Figure 5). A non-contrast computed tomography scan of the face was obtained, revealing the full extent of the cortical expansions, detailing the well-defined multilocular lesions and no rupture of the cortical bones (Figures 6 and 7).

Image 5: Panoramic radiography.



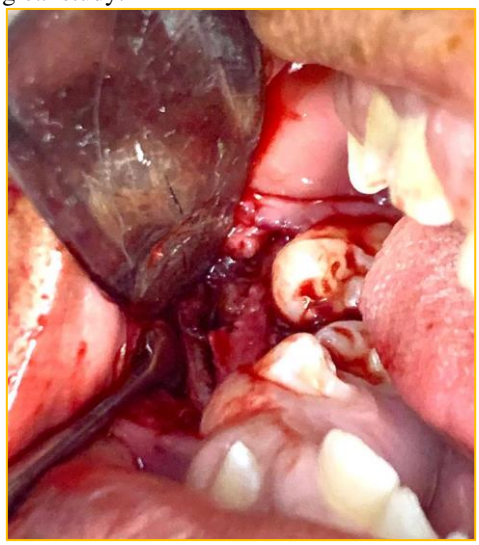
Images 6 and 7: Non-contrast computed tomography of the face in 3D (A), sagittal (B), coronal (C) and axial (D) planes, respectively. Note the dimensions of height and width of the lesion on the right side of the body and more prominent mandibular ramus when compared to the left side



Laboratory investigation was performed, and the blood profile as well as her serum levels of phosphorus 4.9 mg/dL, calcium 9.1 mg/dL, alkaline phosphatase 178 U/I, and parathyroid hormone (PTH) 37.48 mg/dL were within normal limits.

Small fragments of the lesion were obtained through incisional biopsy under local anesthesia with access to the right mandibular vestibule for histopathological evaluation (Figures 8 and 9). A softened bone aspect of purplish color was observed, which was easy to remove from the specimen. The anatomopathological report revealed numerous monomorphonuclear cells, ovoid or spindle-shaped, interspersed with multinucleated giant cells. To perform the surgical procedure, the mother signed the free and informed consent form, authorizing the performance of the same.

Images 8 and 9: Incisional biopsy with access to the right mandibular vestibule fundus. Material collected for anatomopathological study.



Due to limited resources, it was not possible to perform genetic testing. However, the clinical, imaging, and histopathological findings pointed to a non-hereditary presentation of cherubism, and thus the final diagnosis was made. Due to the self-limiting behavior of the pathology, surgical interventions were not necessary and the patient will be followed up in regular evaluations until they reach puberty.

DISCUSSION

Cherubism is a rare autosomal dominant inherited disease, typically manifested as a symmetrical, bilateral, multilocular-looking expansion of the mandible or maxilla affecting children between 2-7 years of age. Despite being considered a genetic disease, some non-hereditary cases have been reported in the literature. Due to the few documented cases, it is difficult to determine the prevalence of this disease [1, 3, 4, 6, 7]. Although most cases presented a symmetrical increase in volume, in this case there was a greater prominence of the right side.

The first manifestations of the conditions usually occur from the age of 2, and vary in severity, and may affect vision, speech, breathing and swallowing in more severe cases [1, 6]. Clinically, a progressive increase in facial volume is observed bilaterally until puberty, when progression stops. Radiographically, the lesion is characteristically multiloculated, radiolucent, well-defined, and symmetrical, with a predilection for the mandible [2, 3]. The non-involvement of the condyles seems to be a patagomonic sign of this condition [3, 5, 8], and it was also one of the findings found in our case, as well as the involvement of only the mandible.

When the maxilla is affected, involvement of the orbit may occur, displacing the eyeballs upwards and the lower eyelid downwards, exposing the sclera and giving the appearance of "eyes turned to the sky" [1]. Such a finding was not present in this patient, and is rarely found in other case reports [6]. In this case, the patient had an asymptomatic increase in volume, without detriment to his other functions.

The lesion can cause tooth malpositioning and lead to eruption disorders, ectopic eruptions, impactions, and malocclusions 3,5. In this case, it was possible to observe impactions caused by the increase in bone and gingival volume and malpositioning of teeth in the arch. The patient did not present caries clinically or radiographically. Periodic assessments of dental condition were encouraged. In the future, if orthodontic intervention is necessary, it should be done after the regression of condition [3,] however, minor surgeries such as extractions and biopsies can be performed without causing progression of the lesions [5].

The diagnosis of cherubism should be made after an in-depth evaluation of the clinical and radiographic findings, family history, laboratory and histopathological analysis [1, 7]. The histological findings of this condition are common to several other bone pathologies, such as the central giant cell lesion and the brown tumor of hyperparathyroidism, which makes the analysis alone insufficient for the final diagnosis of the lesion. The tissue shows multinucleated giant cells in a vascularized stroma of fibrous connective [tissue4,6]. Although this case corroborates this finding, radiographic, clinical, and laboratory findings were essential for diagnostic confirmation.

In laboratory tests, the levels of calcium, phosphorus and PTH, typical markers of bone metabolism, are generally within normal limits in cherubism, although alkaline phosphatase levels may be elevated [1, 4]. The patient in question had borderline levels of calcium, PTH and phosphorus, but had slightly increased alkaline phosphatase levels, corroborating the literature. However, it is not known if this is related to the injury or a possible difficulty in eating properly.

Based on the genetic mutations associated with the disease, genetic testing for the SH3BP2 gene can help confirm the diagnosis of the condition, even in cases without a prior family history. Referral to a medical geneticist can be recommended to parents, so that their doubts can be resolved in a more appropriate way [1, 6]. In our case, due to limitations associated with socioeconomic and structural issues, genetic tests could not be performed on the patient and his parents. Due to these limitations, it was not possible to determine whether the case is a true sporadic case, new genetic mutation or incomplete penetrance of the gene.

Since the lesions have a tendency to spontaneous regression, it is recommended that surgical interventions be postponed until after puberty [2, 3]. However, facial dysmorphia caused by cherubism can affect both the function and aesthetics of the patient, causing emotional and social problems that should not be overlooked in children, being a parameter of indication for surgical interventions in severe cases [7]. After bone maturation, the lesion is filled with granular bone, and conservative cosmetic surgery may be necessary to correct the bone contour [3].

CONCLUSIONS

The diagnosis of the disease should be based mainly on clinical and imaging findings, since histological analysis is of limited value. The dental anomalies associated with the condition make dental follow-up extremely important. There are no defined protocols for the management of cherubism. However, there is a general consensus that surgical interventions, if necessary for aesthetic and/or functional reasons, should be postponed until after puberty.

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