

Cherubism in a Male Adolescent: A Case Report

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Abstract

Background: Cherubism is a rare autosomal dominant inheritance benign disease. A mutation of the SH3BP2 gene from chromosome 4p16.3 has been found in 75% of the reported cases. Other genetic disorders such as Fragile X syndrome, Noonan syndrome and Ramon syndrome have also been found combined with Cherubism presentations. There is a penetrance of 100% among males and 50-70% among females. Bone osteoclastic reabsorption and subsequent fibrous tissue and cyst formation is the primary characteristic of this condition. The objective of this paper is to describe a case of Cherubism in a male adolescent patient with no other comorbidities.

Materials and Methods: A descriptive analysis of a 12-year-old male patient was carried out. Clinicopathological and imaging data were retrieved from patient' medical files from the Dentistry and Radiology and Diagnostic Imaging services of the Federal University Hospital of the Federal University of Espirito Santo.

Results: The patient underwent plain radiographies and Computerized Tomography (CT) evaluation, due to a progressive, painless, bilateral enlargement of the mandible, that was first noted by their parents at four years of age. Radiographies and CT scan of the sinuses demonstrated multiple expansile osteolytic lesions, with imprecise limits, diffusely affecting the mandible, maxilla, the petrous part of the temporal bones and the orbital floors.

Conclusion: Cherubism is a fibro-osseous genetic disorder, characterized by mandible and maxilla bone reabsorption, with subsequent replacement by fibrous tissue. The main involvement occurs at childhood and regression occurs during puberty with some possible remaining deformities, usually mild. Proper diagnose can be made thru clinical and imaging data, with characteristic genetic abnormalities being observed in 75% of the patients.

Keywords: *Cherubism; Radiology; Dentistry; Genetic Syndrome*

Background

Cherubism is a rare autosomal dominant, benign disease. A mutation of the SH3BP2 gene located at the smaller arm of the chromosome 4 at position 16.3 has been found in around 75% of the reported cases [1]. Other genetic disorders such as Fragile X syndrome, Noonan syndrome and Ramon syndrome have also been found combined with Cherubism presentations [2].

There is a penetrance of 100% among males and 50 - 70% among females. Osteoclastic reabsorption of the Mandible and maxilla bones, with subsequent replacement by fibrous tissue, is the main characteristic of this condition. First manifestations are usually observed at the end of the first to the third year of age, a period of time where there is a self-limit bone growing. During puberty the disease ceases and starts to regress, becoming clinically subtle at the end of the third decade of life [3,4].

Jaws and maxilla involvement are symmetric and bilateral, what generates cheek volume increase, especially involving their medial and inferior aspects. This generates a resemblance with renaissance art representation of the Cherubs. Usually it is a pain free condition once it has a slowly progression presentation [3-5].

Histological analysis are inconclusive, once microscopic findings are similar to other conditions such as Giant Cell granuloma, Hyperparathyroidism Brow Tumor, and others. This reinforces the importance to correlate clinical findings and imaging methods to proper define the diagnoses.

Materials and Methods

A descriptive analysis of a 12-year-old male patient was carried out. Clinicopathological and imaging data were retrieved from patient' medical files from the Dentistry and Radiology and Diagnostic Imaging services of the Federal University Hospital of the Federal University of Espirito Santo, Brazil.

Case Report

A 12-year-old male patient that was under outpatient care for 6 months at the dentistry department of a Brazilian Federal Hospital. The major complaint was an progressive, pain free, enlargement of the lower portion of the cheeks for the past 8 years, noted by their parents and family members when palpating the region. There is a family report that a half-brother from his father side has similar facial alterations. His currently complaint is a difficulty to chew some more consistent food, that is attributed to dental arch malformations.

Physical examination reveals facial deformity characterized by bilateral and slightly asymmetrical enlargement of the cheeks, a little more prominent on the right side where a bump of the maxilla is noted at the infraorbital region laterally to the nasal bone (Figure 1). It was not noted proptose, hypertelorism, eye movement disturbances, dysphagia, nor breathing difficulty.



Figure 1: Enlarged symmetrical cheeks and Cherubim-like face.

Plain x-rays revealed multiple expansive osteolytic lesions at the rami and mandible angles, and also maxilla, without losing maxillary sinus pneumatization (Figure 2).

Computerized Tomography scan revealed multiple imprecise limits expansive osteolytic lesions, diffusely observed in the mandible and maxilla (markedly at the tooth alveoli), petrous portion of the temporal bones and orbital floor. It was also observed soft tissue attenuation without significant contrast enhancement, which increases volumetric size associated to mandible and maxilla remodeling. It was not observed condyle nor temporal-mandible joint involvement (Figures 3,4).



Figure 2: Plain films in Coronal and Sagittal planes. Expansive osteolytic lesions at the angles and mandible rami: floating tooth phenomena.

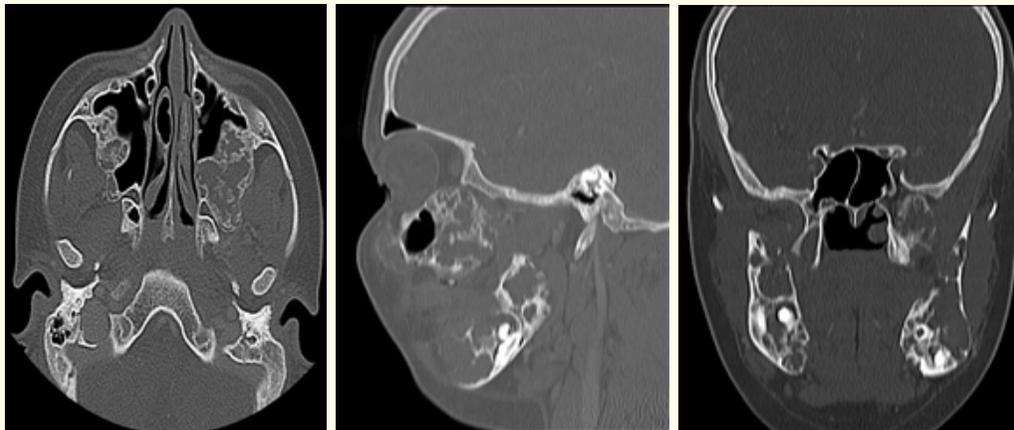


Figure 3: Bone window computerized tomography scan in axial, sagittal and coronal planes. Expansive solid osteolytic lesions at the angles and ascending rami of mandible, maxilla, and petrous portion of the temporal bones.



Figure 4: Coronal and Sagittal Computerized tomography with 3 dimension reconstruction. Expansive solid osteolytic lesions at the angles and ascending rami of mandible, maxilla, and petrous portion of the temporal bones and orbital floor.

Discussion

Cherubism was initially described as an inherited rare family disease, however isolated cases of genetic mutation have also been described [6,7].

Clinical and radiological findings usually start between 14 months-old to three-year-old, when a visible symmetric cheek enlargement is observed. Frequently the disease is more active from 3 to 7 year-old when a higher bone growth is observed, and ceases around the age of 12 to 15 year-old when, at the same time, it starts to regress. Mandible remodeling stays up to the third decade of life when gradually the lesions start to be fulfilled by normal bone. This creates a normal-like facial appearance with only subtle clinical minor changes at the end of this period [5,8].

There is a high phenotype variability, ranging from mild to severe alterations, with a direct correlation with the age of onset. Younger presentations represent more severe deformity cases.

Therefore the signs and symptoms of Cherubism will vary upon severity of presentations, ranging from unnoted minor changes to severe and extensive mandible and maxilla destruction that can even cause respiratory tract obstruction and visual impairment [8].

In more typical patterns, it is noted a mid-lower portion cheek enlargement, skin tension below the eyes with lower eyelid drop, and sclerotic exposure below the iris. These can mimic an upward look of the Cherubim angels as pictured on the Renaissance and Baroque artistic materials. It is also observed tooth and dental arch alterations due to bone coarse remodeling, and tongue dislocations that can be associated with sleeping and breathing disturbances. Despite the variety of anomalies that can be observed in this wide spectrum of presentations, there is no physical or mental development impairment [6]. On the other hand, the same cannot be stated for psychological and social life components that can be deeply affected for lifetime.

Plain film Cherubism presentation is usually composed of bilateral expansive osteolytic mandible lesions, early affecting the posterior body and ascending rami, associated to a coarse trabecular with condyle preservation. Maxilla lesion can simultaneously occur but are not often observed in the early stages of the disease, due to a bone overlapping in this region [5,9].

For a more adequate evaluation of maxilla lesions it is important to use the CT scan, that can reveal more subtle changes with a better resolution that allow a proper characterization of maxilla antro reduction and orbital floor elevation [5,10]. Besides that CT can reveal lesions in a more initial stage, their extension degree [10], tooth alveolar destruction generating the floating tooth phenomena, orbital impairment degrees, and optic nerve compression. Some tooth can be lost or exfoliated prematurely [5,9,11].

Plain films should be used as the first initial imaging method, due to its high sensibility, including in initial disease stages. CT should be used as a subsequent method for a more refined and precise evaluation of soft tissue and bone impairment.

Histopathological study does not have an important role on the diagnosis of Cherubism. Its findings are usually a multinucleate giant cell and fibrous connective tissue, with variable quantities depending upon the disease stage. Older lesions have a higher number of fibrous tissue, lower giant cells, and neo bone formation that correlates to the irregular sclerosis observed when osteolytic areas start to ossify again [7,12].

Treatment modalities are mainly restricted to deformities surgical corrections when indicated, and clinical support [5]. The patient presented here needs to be followed-up for a longer period of time, for a better understanding of his evolution and the need or not of surgical interventions depending upon his physical impairment evolution. However, considering his age and the current disease impairment it is not believed that specific surgical interventions will be needed in the future, and it is expected that it will be improvement up to his third decade of life.

Conclusion

It is clear that the anomalies presented by this patient folds into the diagnose of Cherubism, based only in his clinical and imaging observations (plain films and CT scan), and that histological studies are not usually necessary due to its poor contribution for the diagnostic.

Cherubism is a fibro-osseous genetic disorder, characterized by mandible and maxilla bone reabsorption, with subsequent replacement by fibrous tissue. The main involvement occurs at childhood and regression occurs during puberty with some possible remaining deformities that are usually mild. Proper diagnose can be made thru clinical and imaging data, with characteristic genetic abnormalities being observed in 75% of the patients.

Bibliography

1. Li CY and Yu SF. "A novel mutation in the SH3BP2 gene causes cherubism: case report". *BMC Medical Genetics* 7 (2006): 84.
2. Van Capelle CL, *et al.* "Neurofibromatosis presenting with a cherubism phenotype". *European Journal of Pediatrics* 166.9 (2007): 905-909.
3. Pena N, *et al.* "Querubismo: revisão de literature". *Revista de Ciências Médicas e Biológicas* 3.2 (2004): 261-266.
4. Oliveira FMP, *et al.* "Querubismo: aspectos clínicos, radiográficos e terapêuticos". *Revista do Instituto de Ciências da Saúde* 26.2 (2008): 255-258.
5. Carvalho TN, *et al.* "Relato de caso e revisão da literatura com aspectos imaginológicos". *Radiologia Brasileira* 37.3 (2004): 215-217.
6. Piva F, *et al.* "Querubismo: relato de caso". *Revista Gaucha de Odontologia (Impresso)* 54.3 (2006): 265-268.
7. Ongole R, *et al.* "Cherubism in siblings: a case report". *Journal of the Canadian Dental Association* 69.3 (2003): 150-154.
8. Lannon DA and Earley MJ. "Cherubism and its charlatans". *British Journal of Plastic Surgery* 54.8 (2001): 708-731.
9. Caballero R and Vinals H. "Cherubism: a study of three generations". *Medicina Oral Patologia Oral y Cirugia Bucal* 3.3 (1998): 163-171.
10. Obando FS and Viasus ML. "Querubismo: um caso clínico". *Revista Chilena de Pediatría* 80.1 (2009): 65-69.
11. Ahmadi AJ, *et al.* "Optic neuropathy and macular chorioretinal folds caused by orbital cherubism". *Archives of Ophthalmology* 121.4 (2003): 570-573.
12. Neville BW, *et al.* "Oral and maxillofacial pathology. 2nd edition". Philadelphia, PA: WB Saunders (2002): 547-549.

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