



Cherubism

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ABSTRACT

Cherubism, a very rare disorder with only an estimated 300 cases reported in the literature, is a benign hereditary condition of the mandible and/or maxilla, usually found in children between 2 and 5 years of age giving characteristic cherubic appearance to the patient. On radiography, the lesions exhibit bilateral multilocular radiolucent areas. Histopathology reveals multinucleated giant cells in the background of proliferating fibrous connective tissue. The present case report describes cherubism in a 10 years old male child and briefly reviews literature on this report.

Keywords: Autosomal dominant, Cherubism, Giant cells, Fibrous dysplasia.

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INTRODUCTION

Cherubism is a rare benign, self-enlargement of the mandible and/or maxilla. It was first described in 1933 by Jones,¹ usually found in children between 2 and 5 years of age.² It is a familial disease and presents as autosomal dominant trait with 100% penetrance in males and 50 to 70% penetrance in females.¹ Initially, called as 'familial fibrous dysplasia of the jaws', limiting fibro-osseous genetic disorder of the jaw bones characterized by bilateral, painless, symmetrical but recent genetic investigations have proved it as to be separate entity at the molecular level.³ The word 'cherub' refers to angels with childish full cheeked face (chubby cheeks) often gazing upward as if eyes to heaven as depicted in the Renaissance era.⁴ The patients typically present with marked fullness of the jaws and cheeks with upward gazing of the eyes.² On the basis of extent of involvement, Ramon and Engelberg⁵ proposed a grading system for cherubism: grade 1 (involvement of both

mandibular ascending rami; grade 2 (same as grade 1 plus involvement of both maxillary tuberosities); grade 3 (massive involvement of whole maxilla and mandible, except the condylar processes); grade 4 (same as grade 3 with involvement of the floor of the orbits causing orbital compression). Although extragnathic skeletal involvement is rare,¹ Davis et al⁶ reported some rare occurrences in other bones, i.e. ribs, humerus and femur.

CASE REPORT

A 10 years old male child presented with chief complaints of painless bilateral bony swellings in the lower jaw (Fig. 1). History revealed that the parents of the child observed the swellings on both the sides of lower jaw of their child at the age of 5 years that progressively increased to present size. The child did not have any contributory past and family history. An extraoral clinical examination revealed chubby cheeks with bony hard swellings bilaterally at the angle region of mandible. It was roughly oval in shape, fixed, hard in consistency with ill-defined margins but no secondary changes. Temperature of the overlying skin was normal and no tenderness elicited. Intraorally, mandibular buccal vestibule was obliterated bilaterally posterior to premolars. Blood investigations (Sr. calcium, phosphorus, alkaline phosphatase) were normal. An orthopantomograph (Fig. 2) showed multilocular radiolucencies involving the ramus and angle regions of the mandible and posterior maxilla bilaterally. Axial CT scan revealed the expansile multilocular radiolucencies at the mandibular ramus and angle bilaterally with intact cortices (Fig. 3). Coronal cuts showed the multilocular radiolucencies involving the maxilla with partial obliteration of antra bilaterally (Fig. 4).

Biopsy was taken under general anesthesia and the histopathological examination showed a highly cellular mature fibrous connective tissue with numerous endothelial cell proliferations along with multinucleated giant cells (Fig. 5).

DISCUSSION

According to the World Health Organization classification, cherubism belongs to a group of non-neoplastic bone lesions that affect only the jaws.⁷ Anderson and McCleden reviewed 65 cases from 21 families and suggested that the pattern of inheritance was autosomal dominant.⁸

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Fig. 1: Characteristic appearance of cherubic patient

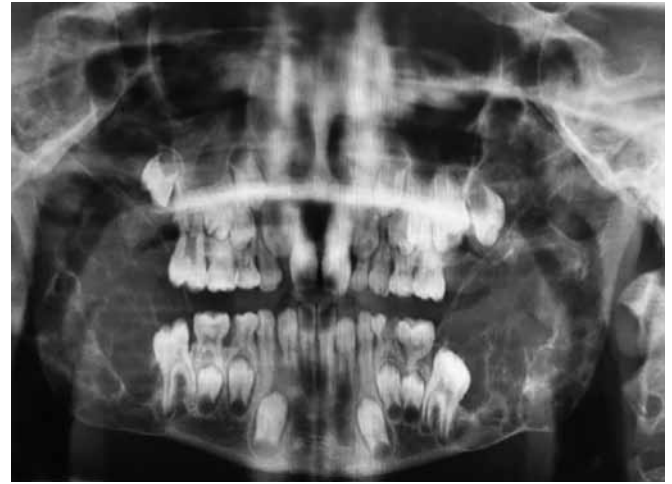


Fig. 2: OPG of the patient showing bilateral multilocular radiolucencies involving the angle, ramus and coronoid processes of the mandible as well as the maxillary tuberosities bilaterally. The condyle is characteristically spared bilaterally

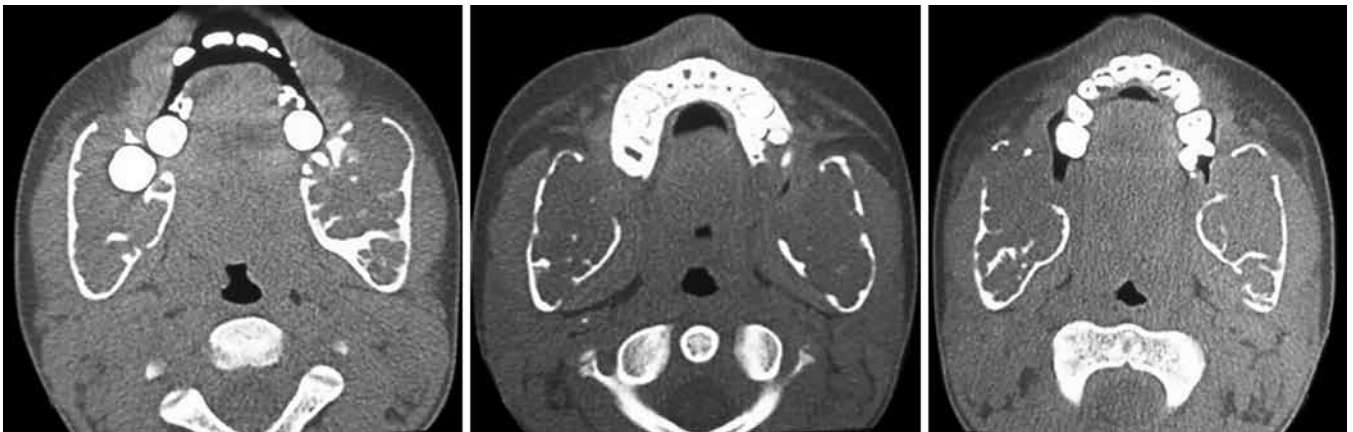


Fig. 3: Axial CT reveals multilocular radiolucencies with cortical expansion at mandibular ramus as well as maxillary tuberosity regions bilaterally



Fig. 4: Coronal cuts showing obliteration of maxillary antra with the expanding mass bilaterally

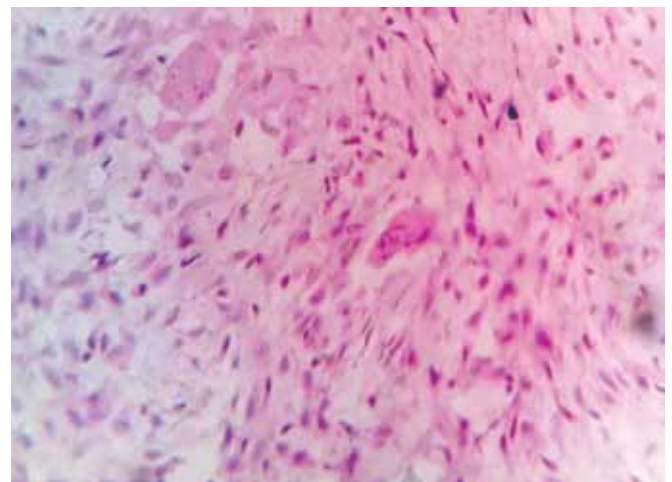


Fig. 5: Photomicrograph of the biopsy specimen

Sporadic cases have also been reported. Patients typically present with a painless symmetric enlargement of the posterior region of the mandible which achieves considerable size before it is detected, and it can produce

severe facial deformity.⁹ The first signs of manifestation of the disease are generally observed at about 2 years of age, followed by accelerated growth from 8 to 9 years and spontaneous interruption after puberty.¹⁰ The

phenotype ranges from no clinical manifestation to severe mandibular and maxillary overgrowth with respiratory, vision, speech and swallowing problems. Intraorally, it presents as a hard, nontender swelling palpable in the affected area. Sub-mandibular and upper cervical lymphadenopathy² are common, although reactive regional lymphadenopathy, particularly of the sub-mandibular lymph nodes, usually subsides after 5 years of age. Cherubism is reported to be associated with some well described syndromes, including neurofibromatosis type 1, Noonan-like/multiple giant cell lesion syndrome, Ramon syndrome, and Jaffe-Campanacci syndrome.⁴

Radiographically, it appears as numerous well-defined multilocular radiolucencies of the jaws. The borders are distinct and divided by bony trabeculae. In mandible, it causes thinning and expansion of the cortical plates with occasional perforation. Displacement of the inferior alveolar canal may be noted. Sparing of mandibular condyles was earlier considered a hallmark of this condition; however, condylar involvement has also been described.¹¹ Unerupted teeth are often displaced and appear to be floating in the cyst like spaces.

Marck and Kudryk¹² reported that conventional radiography provided a limited image because it is only two dimensional. On the other hand, CT provided a realistic picture of the lesions, showing some aspects that otherwise would not be demonstrable due to superimposition and the anatomical complexity of the jaws. Additionally, the 3D formatting contributes to the diagnosis.¹³ The MRI findings of cherubism were first described by Beaman et al, who described cherubic lesions as nonspecific, homogeneous, isointensity to skeletal muscle on T1W images and heterogeneous isointensity on fat suppressed T2W images.¹⁰

Mnari et al reported that MRI is helpful for determining soft tissue involvement in patients with aggressive cherubism and for assessing the vascular structures pre-operatively.⁴

Histologically, the lesions are composed of a vascularized fibrous stroma containing multinucleated giant cells, resembling giant cell granuloma.² The genetic basis for cherubism was identified in 1999, when the gene responsible for it was mapped to chromosome 4p16.3.¹⁴ Mutation of the gene encoding for fibroblast growth factor receptor III (FGF-RIII) has also been found in some cases of cherubism.¹⁵ Histopathologically, the lesion has been characterized into three subtypes: I—predominance of multinuclear cells; II—predominance of inflammatory activity; and III—predominance of fibrosis.

As the patient grows, the jaw bone tends to resolve and progressively resulting in a normal jaw configuration.¹⁴

In addition, several authors have reported that in spite of being a self-limiting condition that subsides with age, it can cause serious orbital abnormalities and impairment of hearing.⁹ Being a self-limiting condition, treatment is mainly for the esthetic needs and for unerupted teeth. Liposuction has been proposed to reduce the mass of the lesion in particular cases.¹ Curettage alone or in combination with surgical contouring for cosmetic purposes has been considered the treatment of choice and some authors have reported a massive growth of the lesion after surgery. Some authors point medical therapy in the form of calcitonin as a possibility to curtail the disease and obviate the need for surgery, but only a few references are available¹⁵ in the literature.

CONCLUSION

Cherubism is a rare osseous disorder of children and adolescents. Although the radiologic characteristics of cherubism are not pathognomic, the diagnosis is strongly suggested by bilateral, relatively symmetric jaw involvement that is limited to maxilla and mandible. Imaging typically shows expansile remodeling of involved bones, thinning of cortexes and multilocular radiolucencies with coarse trabecular pattern. Nowadays, genetic tests should be used for the final diagnosis of cherubism. Being a self-regressing condition, generally, minimally invasive treatment is done; however, sometimes surgical management is required for cosmetic reasons.¹⁶

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