

Cherubism- A Case Report with Review

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

Index Terms- Autosomal dominant, cherubism, giant cells, fibrous dysplasia, OPG

I. INTRODUCTION

Cherubism, first described by Jones,¹ is a benign, self-limiting fibro-osseous disorder characterized by bilateral expansion of the mandible, maxilla, or both. The lesions are usually symmetrical and painless. Cherubism is a benign hereditary condition of the jaws, usually found in children by 5 years of age.² It is a familial disease and presents an autosomal dominant trait with 100% penetrance in males and 50–70% penetrance in females.¹ Initially called as ‘familial fibrous dysplasia of the jaws’, 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 gaizing upwards 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 extra-gnathic skeletal involvement is rare, 1 Davis et al⁶ reported some rare occurrences in other bones, i.e., ribs, humerus, and femur. The authors in this article present a case report of a 10-years old male patient with swelling on both sides of the upper and lower jaw since two years after birth.

II. CASE REPORT

A 10-years old male child presented with chief complaints of bilateral swellings in the both jaws and bilateral bony

enlargement in the body of cheek without any pain. History revealed that it started as a small swelling since 2 years that progressively increased to present size. The child did not have any contributory past and family history. An extra-oral clinical examination showed a diffuse enlargement of maxillary region and bilaterally at the angle of mandible. It was roughly oval in shape, hard in consistency with ill-defined margins. The swelling was fixed with no secondary changes. Swelling on the right side was slightly larger(Figure 1&2). On palpation, temperature of the overlying skin was normal and no tenderness elicited. Intra-orally, there was a visible diffuse swelling present at both upper and lower jaw. Dental examination was normal. There was no syndromic association. Blood investigations (Sr. calcium, phosphorus, alkaline phosphatase) were normal. An orthopantomograph(Figure 3) and x-ray paranasal sinus Water’s & Caldwell’s view of the patient showed multilocular appearance of the mandible and maxilla(Figure 4&5). Histopathological examination showed a highly cellular mature fibrous connective tissue with numerous endothelial cell proliferations along with multinucleated giant cells. (Figure 6)

Figure 1&2: extra-oral clinical examination showing a diffuse enlargement of maxillary region and bilaterally at the angle of mandible





Figure3: OPG showing expansile multilocular bilateral spaces in both mandible and maxilla. Cortex is thinned out with intact margins. Displaced and deranged upper and lower teeth; both erupted and unerupted.

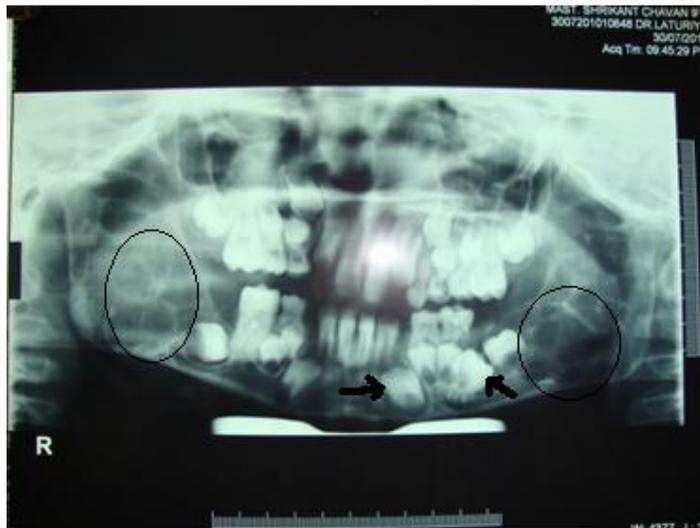


Figure 4&5: x-rays Paranasal sinus showing multilocular spaces of cherubism

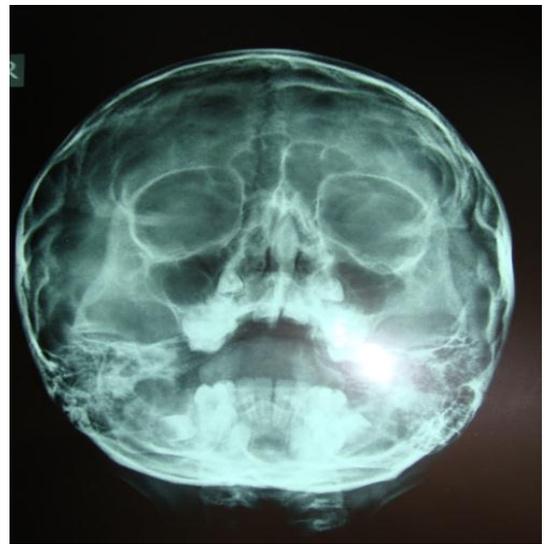
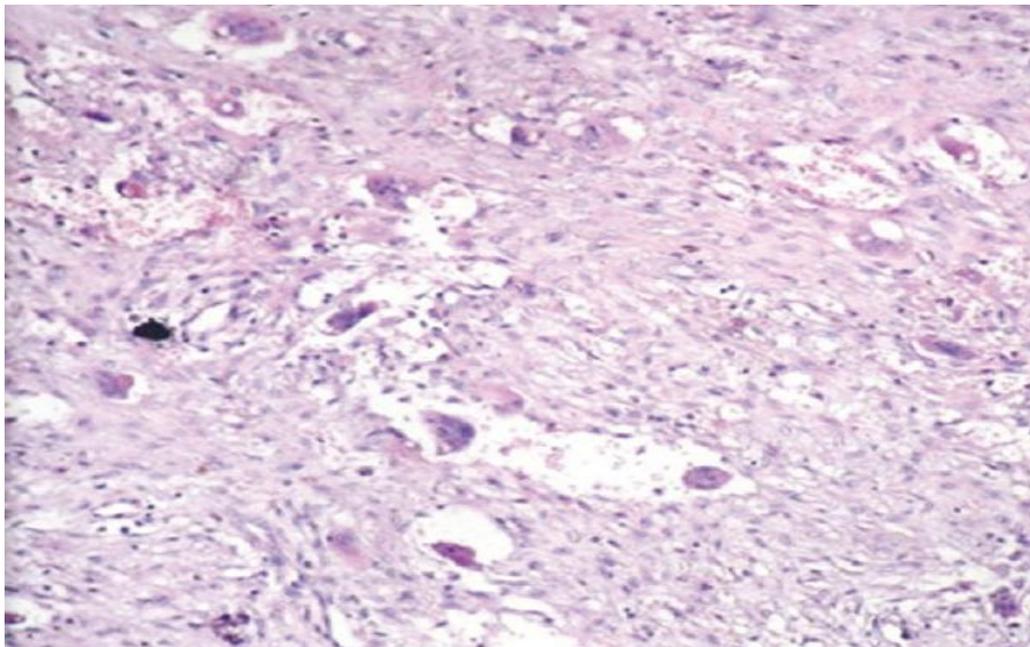


Figure 6: Histopathological picture showing the multinucleated giant cells with pre-vascular eosinophilic cuffing.



III. 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 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. Intra-orally it presents as a hard, non-tender swelling palpable in the affected area. Sub-mandibular and upper cervical lymphadenopathy² are common, although reactive regional lymphadenopathy, particularly of the submandibular 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 3-D 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 iso-intensity 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.

IV. 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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